

STN Columbus

* * * * * Welcome to STN International * * * * *

NEWS	1		Web Page for STN Seminar Schedule - N. America
NEWS	2	AUG 06	CAS REGISTRY enhanced with new experimental property tags
NEWS	3	AUG 06	FSTA enhanced with new thesaurus edition
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NEWS	5	AUG 20	CA/Capplus enhanced with CAS indexing in pre-1907 records
NEWS	6	AUG 27	Full-text patent databases enhanced with predefined patent family display formats from INPADOCDB
NEWS	7	AUG 27	USPATOLD now available on STN
NEWS	8	AUG 28	CAS REGISTRY enhanced with additional experimental spectral property data
NEWS	9	SEP 07	STN AnaVist, Version 2.0, now available with Derwent World Patents Index
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NEWS	11	SEP 13	INPADOCDB enhanced with monthly SDI frequency
NEWS	12	SEP 17	CA/Capplus enhanced with printed CA page images from 1967-1998
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NEWS	16	OCT 19	BEILSTEIN updated with new compounds
NEWS	17	NOV 15	Derwent Indian patent publication number format enhanced
NEWS	18	NOV 19	WPIX enhanced with XML display format
NEWS	19	NOV 30	ICSD reloaded with enhancements
NEWS	20	DEC 04	LINPADOCDB now available on STN
NEWS	21	DEC 14	BEILSTEIN pricing structure to change
NEWS	22	DEC 17	USPATOLD added to additional database clusters
NEWS	23	DEC 17	IMSDRUGCONF removed from database clusters and STN
NEWS	24	DEC 17	DGENE now includes more than 10 million sequences
NEWS	25	DEC 17	TOXCENTER enhanced with 2008 MeSH vocabulary in MEDLINE segment
NEWS	26	DEC 17	MEDLINE and LMEDLINE updated with 2008 MeSH vocabulary
NEWS	27	DEC 17	CA/Capplus enhanced with new custom IPC display formats
NEWS	28	DEC 17	STN Viewer enhanced with full-text patent content from USPATOLD
NEWS	29	JAN 02	STN pricing information for 2008 now available
NEWS	30	JAN 16	CAS patent coverage enhanced to include exemplified prophetic substances
NEWS	31	JAN 28	USPATFULL, USPAT2, and USPATOLD enhanced with new custom IPC display formats
NEWS	32	JAN 28	MARPAT searching enhanced
NEWS	33	JAN 28	USGENE now provides USPTO sequence data within 3 days of publication
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NEWS	35	JAN 28	MEDLINE and LMEDLINE reloaded with enhancements
NEWS	36	FEB 08	STN Express, Version 8.3, now available

NEWS EXPRESS FEBRUARY 08 CURRENT WINDOWS VERSION IS V8.3,
AND CURRENT DISCOVER FILE IS DATED 24 JANUARY 2008

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NEWS LOGIN Welcome Banner and News Items
NEWS IPC8 For general information regarding STN implementation of IPC 8

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FILE 'HOME' ENTERED AT 13:50:30 ON 19 FEB 2008

=> file medline caplus
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=> s SNP and database and (disease or drug) and (identify or scan or specify or target)
L1 222 SNP AND DATABASE AND (DISEASE OR DRUG) AND (IDENTIFY OR SCAN OR SPECIFY OR TARGET)

=> s l1 and haplotype
L2 55 L1 AND HAPLOTYPE

=> dup rem l2
PROCESSING COMPLETED FOR L2
L3 41 DUP REM L2 (14 DUPLICATES REMOVED)

=> d bib ab 1-41

L3 ANSWER 1 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2007:590726 CAPLUS

DN 146:521186

TI System and method for genotyping single nucleotide polymorphisms associated with genetic **diseases** and susceptibility for therapy and using genetic, phentotypic and clinical data to make predictions

IN Rabinowitz, Matthew; Banjevic, Milena; Demko, Zachary Paul; Johnson, David Scott

PA Gene Security Network LLC, USA

SO PCT Int. Appl., 163pp.

CODEN: PIXXD2

DT Patent

LA English

FAN.CNT 2

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	WO 2007062164	A2	20070531	WO 2006-US45281	20061122
	WO 2007062164	A3	20071129		
	W:	AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IS, JP, KE, KG, KM, KN, KP, KR, KZ, LA, LC, LK, LR, LS, LT, LU, LV, LY, MA, MD, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RS, RU, SC, SD, SE, SG, SK, SL, SM, SV, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW			
	RW:	AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IS, IT, LT, LU, LV, MC, NL, PL, PT, RO, SE, SI, SK, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG, BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM, AP, EA, EP, OA			
	US 2007027636	A1	20070201	US 2006-496982	20060731
	US 2007184467	A1	20070809	US 2006-603406	20061122
	US 2007178501	A1	20070802	US 2006-634550	20061206
PRAI	US 2005-739882P	P	20051126		
	US 2005-742305P	P	20051206		
	US 2005-754396P	P	20051229		
	US 2006-774976P	P	20060221		
	US 2006-789506P	P	20060404		
	US 2006-817741P	P	20060630		
	US 2006-496982	A	20060731		

US 2006-846610P P 20060922
 US 2005-703415P P 20050729
 US 2006-846589P P 20060922

AB The present invention provides systems and methods for genotyping single nucleotide polymorphisms in genes and predicting likely phenotypic outcomes using math. models, given genetic, phenotypic and/or clin. data of an individual, and also relevant aggregated medical data consisting of genotypic, phenotypic, and/or clin. data from germane patient subpopulations. Genetic data for the **target** individual is acquired and amplified using known methods, and poorly measured base pairs, missing alleles and missing regions are reconstructed using expected similarities between the **target** genome and the genome of genetically related subjects. In one embodiment of the invention, incomplete genetic data from an embryonic cell is reconstructed using the more complete genetic data from a larger sample of diploid cells from one or both parents, with or without genetic data from haploid cells from one or both parents, and/or genetic data taken from other related individuals. In another embodiment, incomplete genetic data from a fetus is acquired from fetal cells, or cell-free fetal DNA isolated from the mother's blood, and the incomplete genetic data is reconstructed using the more complete genetic data from a larger sample diploid cells from one or both parents, with or without genetic data from haploid cells from one or both parents, and/or genetic data taken from other related individuals. In another embodiment, the genetic data can be reconstructed for the purposes of making phenotypic predictions. In another embodiment, the genetic data can be used to detect for aneuploidies and uniparental disomy. In another embodiment, phenotypic predictions may be made using models based on contingency tables for genetic data that can be constructed from data available in genomic **databases**. In another embodiment, a plurality of models are created and tested using a set of test data, and the prediction is made using the model that is identified as the most accurate.

L3 ANSWER 2 OF 41 MEDLINE on STN DUPLICATE 1

Full Text

AN 2007301660 MEDLINE

DN PubMed ID: 17512363

TI Multiple less common genetic variants explain the association of the cholesteryl ester transfer protein gene with coronary artery **disease**.

AU Horne Benjamin D; Camp Nicola J; Anderson Jeffrey L; Mower Chrissa P; Clarke Jessica L; Kolek Matthew J; Carlquist John F

CS Cardiovascular Department, LDS Hospital, Intermountain Medical Center, University of Utah, Salt Lake City, Utah 84143, USA. (Intermountain Heart Collaborative Study Group). benjamin.horne@intermountainmail.org

NC CA098364 (United States NCI)
 CA099844 (United States NCI)
 HL071878 (United States NHLBI)
 HL073117 (United States NHLBI)

SO Journal of the American College of Cardiology, (2007 May 22) Vol. 49, No. 20, pp. 2053-60. Electronic Publication: 2007-05-04.
 Journal code: 8301365. E-ISSN: 1558-3597.

CY United States

DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)

LA English

FS Abridged Index Medicus Journals; Priority Journals

FS CLINICALTRIALS.GOV

NCT NCT00406185

EM 200706

ED Entered STN: 22 May 2007

Last Updated on STN: 28 Jun 2007

Entered Medline: 27 Jun 2007

AB **OBJECTIVES:** The objective of this study was to **identify** associations of the cholesteryl ester transfer protein (CETP) gene with coronary artery **disease** (CAD) with tagging (t) single nucleotide polymorphisms (**SNPs**) chosen to optimally account for intra-genic variation. **BACKGROUND:** The CETP gene plays a critical role in lipoprotein metabolism, but the common and well-studied TaqIB variant is inconsistently predictive of CAD. **METHODS:** From a deoxyribonucleic acid bank of 10,020 individuals, nondiabetic nonsmoking patients (n = 4,811) with angiographically defined, clinically significant CAD (> or =70% stenosis) or normal coronaries were genotyped for 11 CETP tSNPs. Myocardial infarction (MI) and lipid levels

were evaluated as secondary end points. RESULTS: Analysis of single tSNPs, corrected for multiple comparisons ($p < 0.00485$), identified allele +1086A to be associated with CAD ($p = 0.0034$). Suggestive allelic and significant genotypic associations were found for -631AA (odds ratio [OR] = 3.95, $p = 0.004$ vs. CC) and +2389GA (OR = 1.21, $p = 0.003$ vs. GG). **Haplotype** analysis by linkage disequilibrium (LD) group revealed a CAD association for LD group B ($p = 0.0025$ across T+1086A, C+878T, C+408T) and near significance for LD group A ($p = 0.013$ across C-631A, MspI, G+2389A). A weak protective trend for TaqIB was eliminated by adjustment for other tSNPs, and **haplotype** analyses suggested that TaqIB was simply a marker for other tSNPs or **haplotypes**. No tSNP or **haplotype** associations with MI were found. CONCLUSIONS: Multiple, less common **SNPs** and **haplotype** variants underlie CETP-related CAD risk, for which the common TaqIB variant is simply a poor marker. The occurrence of risk-related variants on separate **haplotypes** suggests genetic-risk complexity and allelic heterogeneity. (Database Registry of the Intermountain Heart Collaborative Study; <http://clinicaltrials.gov/ct/show/NCT00406185?order=1;NCT00406185>).

L3 ANSWER 3 OF 41 MEDLINE on STN DUPLICATE 2
Full Text
 AN 2007295209 MEDLINE
 DN PubMed ID: 17412754
 TI Signatures of recent positive selection at the ATP-binding cassette **drug** transporter superfamily gene loci.
 AU Wang Zihua; Wang Jingbo; Tantoso Erwin; Wang Baoshuang; Tai Amy Y P; Ooi London L P J; Chong Samuel S; Lee Caroline G L
 CS Department of Biochemistry, Yong Loo Lin School of Medicine, National University of Singapore, Singapore.
 SO Human molecular genetics, (2007 Jun 1) Vol. 16, No. 11, pp. 1367-80. Electronic Publication: 2007-04-05. Journal code: 9208958. ISSN: 0964-6906.
 CY England: United Kingdom
 DT (COMPARATIVE STUDY)
 Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 (VALIDATION STUDIES)
 LA English
 FS Priority Journals
 EM 200709
 ED Entered STN: 18 May 2007
 Last Updated on STN: 28 Sep 2007
 Entered Medline: 27 Sep 2007
 AB Members of the ATP-binding cassette (ABC) superfamily of transporters have been implicated as major players in **drug** response. Single nucleotide polymorphisms (**SNPs**) in the ABC transporter genes may account for variation in **drug** response between individuals. Given the abundance of **SNPs** within the human genome, identification of functionally important **SNPs** is difficult. Here, we utilized signatures of recent positive selection (RPS) to **identify SNPs** in ABC genes that have potential functional significance by using the long-range-**haplotype** test to search for signatures of RPS at 18 ABC genes involved in **drug** transport. From the genotype data of these 18 ABC genes in four populations extracted from the HapMap **database**, at least one **SNP** in each of these genes displayed genomic signatures of RPS in at least one population. However, only 13 **SNPs** in 10 ABC genes from three populations retained statistical significance after Type I error reduction. The functional significance of six of these RPS **SNPs**, including those that failed multiple testing correction (MTC), has been reported previously. We experimentally confirmed a functional effect for two **SNPs**, including one that failed to show evidence of RPS after MTC. These observations suggest that Type I error reduction may inadvertently increase Type II error. Although the remaining positively selected **SNPs** have yet to be functionally validated, our study illustrates the feasibility of using this strategy to **identify SNPs** within 'adaptive' genes that may confer functional effect, prior to testing their roles in individual/population **drug** response variation or in complex **disease** susceptibility.

L3 ANSWER 4 OF 41 MEDLINE on STN
Full Text
 AN 2007488866 MEDLINE
 DN PubMed ID: 17597650

TI Comprehensive genetic variant discovery in the surfactant protein B gene.
 AU Hamvas Aaron; Wegner Daniel J; Carlson Christopher S; Bergmann Kelly R;
 Trusgnich Michelle A; Fulton Lucinda; Kasai Yumi; An Ping; Mardis Elaine
 R; Wilson Richard K; Cole F Sessions
 CS Edward Mallinckrodt Department of Pediatrics, Washington University School
 of Medicine and St. Louis Children's Hospital, St. Louis, Missouri 63110,
 USA.
 NC R01 HL 065174 (United States NHLBI)
 R01 HL 065385 (United States NHLBI)
 U54 HG 003079 (United States NHGRI)
 SO Pediatric research, (2007 Aug) Vol. 62, No. 2, pp. 170-5.
 Journal code: 0100714. ISSN: 0031-3998.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 LA English
 FS Priority Journals
 EM 200709
 ED Entered STN: 22 Aug 2007
 Last Updated on STN: 20 Sep 2007
 Entered Medline: 19 Sep 2007
 AB Completely penetrant mutations in the surfactant protein B gene (SFTPB)
 and >75% reduction of SFTPB expression disrupt pulmonary surfactant
 function and cause neonatal respiratory distress syndrome. To inform
 studies of genetic regulation of SFTPB expression, we created a catalogue
 of SFTPB variants by comprehensive resequencing from an unselected,
 population-based cohort (n = 1,116). We found an excess of low-frequency
 variation [81 **SNPs** and five small insertion/deletions (in/dels)].
 Despite its small genomic size (9.7 kb), SFTPB was characterized by weak
 linkage disequilibrium (LD) and high **haplotype** diversity. Using the
 HapMap Yoruban and European populations, we identified a recombination hot
 spot that spans SFTPB, was not detectable in our focused resequencing
 data, and accounts for weak LD. Using homology-based software tools, we
 discovered no definitively damaging exonic variants. We conclude that
 excess low-frequency variation, intragenic recombination and lack of
 common disruptive exonic variants favor complete resequencing as the
 optimal approach for genetic association studies to **identify** regulatory
 SFTPB variants that cause neonatal respiratory distress syndrome in
 genetically diverse populations.

L3 ANSWER 5 OF 41 MEDLINE on STN
Full Text
 AN 2007397747 MEDLINE
 DN PubMed ID: 17446335
 TI The SERPINE2 gene is associated with chronic obstructive pulmonary
disease in two large populations.
 AU Zhu Guohua; Warren Liling; Aponte Jennifer; Gulsvik Amund; Bakke Per;
 Anderson Wayne H; Lomas David A; Silverman Edwin K; Pillai Sreekumar G
 CS GlaxoSmithKline R&D, 5 Moore Drive, Research Triangle Park, NC 27709, USA.
 (International COPD Genetics Network (ICGN) Investigators).
 SO American journal of respiratory and critical care medicine, (2007 Jul 15)
 Vol. 176, No. 2, pp. 167-73. Electronic Publication: 2007-04-19.
 Journal code: 9421642. ISSN: 1073-449X.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Abridged Index Medicus Journals; Priority Journals
 EM 200709
 ED Entered STN: 10 Jul 2007
 Last Updated on STN: 19 Sep 2007
 Entered Medline: 18 Sep 2007
 AB RATIONALE: Chronic obstructive pulmonary **disease** (COPD) is a complex
disease influenced by multiple genes and environmental factors. A
 region on chromosome 2q has been shown to be linked to COPD. A positional
 candidate gene from the chromosome 2q region SERPINE2 (Serpine peptidase
 inhibitor, clade E [nexin, plasminogen activator inhibitor type 1], member
 2), was previously evaluated as a susceptibility gene for COPD in two
 association studies, but the results were contradictory. OBJECTIVES: To
identify the relationship between SERPINE2 polymorphisms and
 COPD-related phenotypes using family-based and case-control association
 studies. METHODS: In the present study, we genotyped 25 single nucleotide

polymorphisms (**SNPs**) from SERPINE2 and analyzed qualitative and quantitative COPD phenotypes in 635 pedigrees with 1,910 individuals and an independent case-control population that included 973 COPD cases and 956 control subjects. The family data were analyzed using family-based association tests. The case-control data were analyzed using logistic regression and linear models. MEASUREMENTS AND MAIN RESULTS: Six **SNPs** demonstrated significant associations with COPD phenotypes in the family-based association analysis ($0.0016 \leq p \leq 0.042$). Five of these **SNPs** demonstrated replicated associations in the case-control analysis ($0.021 \leq p \leq 0.031$). In addition, the results of **haplotype** analyses supported the results from single **SNP** analyses. CONCLUSIONS: These data provide further support for SERPINE2 as a COPD susceptibility gene.

L3 ANSWER 6 OF 41 MEDLINE on STN DUPLICATE 3
Full Text
 AN 2007527023 MEDLINE
 DN PubMed ID: 17689071
 TI Human 3beta-hydroxysteroid dehydrogenase types 1 and 2: Gene sequence variation and functional genomics.
 AU Wang Liewei; Salavaggione Ezequiel; Pelleymounter Linda; Eckloff Bruce; Wieben Eric; Weinshilboum Richard
 CS Division of Clinical Pharmacology, Department of Molecular Pharmacology and Experimental Therapeutics, Mayo Clinic College of Medicine, 200 First Street SW, Rochester, MN 55905, USA.. wang.liewei@mayo.edu
 NC R01 GM28157 (United States NIGMS)
 R01 GM35720 (United States NIGMS)
 U01 GM61388 (United States NIGMS)
 SO The Journal of steroid biochemistry and molecular biology, (2007 Oct) Vol. 107, No. 1-2, pp. 88-99. Electronic Publication: 2007-06-08. Journal code: 9015483. ISSN: 0960-0760.
 CY England: United Kingdom
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 LA English
 FS Priority Journals
 EM 200712
 ED Entered STN: 11 Sep 2007
 Last Updated on STN: 28 Dec 2007
 Entered Medline: 27 Dec 2007
 AB The 3beta-hydroxysteroid dehydrogenase/Delta(5)-Delta(4) isomerase isoenzymes 1 and 2 (HSD3B1 and HSD3B2) are membrane-bound enzymes that play essential roles in the biosynthesis of steroid hormones. Therefore, variation in the HSD3B1 and HSD3B2 genes might play a role in the pathophysiology of steroid hormone-related **disease**. We set out to systematically **identify** common polymorphisms and **haplotypes** in human HSD3B1 and HSD3B2. We identified 17 single nucleotide polymorphisms (**SNPs**) in HSD3B1 and 9 in HSD3B2 - the majority of which were not present in public **databases** - by resequencing human HSD3B1 and HSD3B2 using 240 DNA samples from four different ethnic groups (60 samples per group). Functional genomic studies of the five non-synonymous cSNPs in HSD3B1 and the one observed in HSD3B2 showed that two of these polymorphisms resulted in significant decreases in the quantity of enzyme protein expressed. However, none of the three non-synonymous **SNPs** located in areas encoding putative membrane-binding domains altered subcellular localization of the enzyme as determined by immunofluorescence microscopy. Finally, common variant **haplotypes** in the 5'-flanking regions of these genes showed significant cell line-dependent variation in their ability to drive transcription. In aggregate, these results provide a basis for study of the possible role in human **disease** of common genetic variation in HSD3B1 and HSD3B2.

L3 ANSWER 7 OF 41 MEDLINE on STN DUPLICATE 4
Full Text
 AN 2006749214 MEDLINE
 DN PubMed ID: 17172834
 TI Integration of HapMap-based **SNP** pattern analysis and gene expression profiling reveals common **SNP** profiles for cancer therapy outcome predictor genes.
 AU Glinsky Gennadi V
 CS Ordway Cancer Center, Ordway Research Institute, Inc., Albany, New York 12208, USA.. gglinsky@ordwayresearch.org

NC 5R01 CA89827 (United States NCI)
 SO Cell cycle (Georgetown, Tex.), (2006 Nov) Vol. 5, No. 22, pp. 2613-25.
 Electronic Publication: 2006-11-15.
 Journal code: 101137841. E-ISSN: 1551-4005.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 LA English
 FS Priority Journals
 EM 200702
 ED Entered STN: 27 Dec 2006
 Last Updated on STN: 13 Feb 2007
 Entered Medline: 12 Feb 2007
 AB Recent completion of the initial phase of a **haplotype** map of human genome (www.hapmap.org) provides opportunity for integrative analysis on a genome-wide scale of microarray-based gene expression profiling and **SNP** variation patterns for discovery of cancer-causing genes and genetic markers of therapy outcome. Here we applied this approach for analysis of **SNPs** of cancer-associated genes, expression profiles of which predicts the likelihood of treatment failure and death after therapy in patients diagnosed with multiple types of cancer. Unexpectedly, this analysis reveals a common **SNP** pattern for a majority (60 of 74; 81%) of analyzed cancer treatment outcome predictor (CTOP) genes. Our analysis suggests that heritable germ-line genetic variations driven by geographically localized form of natural selection determining population differentiations may have a significant impact on cancer treatment outcome by influencing the individual's gene expression profile. We demonstrate a translational utility of this approach by building a highly informative CTOP algorithm combining prognostic power of multiple gene expression-based CTOP models derived from signatures of oncogenic pathways associated with activation of BMI1; Myc; Her2/neu; Ras; beta-catenin; Suz12; E2F; and CCND1 oncogenes. Application of a CTOP algorithm to large **databases** of early-stage breast and prostate tumors **identifies** cancer patients with 100% probability of a cure with existing cancer therapies as well as patients with nearly 100% likelihood of treatment failure, thus providing a clinically feasible framework essential for introduction of rational evidence-based individualized therapy selection and prescription protocols. Our analysis indicates that genetic determinants of human **disease** susceptibility and severity are encoded by population differentiation **SNP** variants. Evolution of these **SNPs** is driven by geographically-localized form of natural selection causing population differentiation. Recent analysis **identifies** a class of **SNPs** regulating gene expression in normal individuals and likely determining unique genome-wide expression profiles of each individual. We propose that critical **disease**-causing combinations of **SNP** variants arise from **SNPs** regulating mRNA levels and determining genome-wide **haplotype** patterns of individual's **disease** susceptibility.

L3 ANSWER 8 OF 41 MEDLINE on STN DUPLICATE 5
Full Text
 AN 2006516765 MEDLINE
 DN PubMed ID: 16865697
 TI Ethnic variation in AMD-associated complement factor H polymorphism p.Tyr402His.
 AU Grassi Michael A; Fingert John H; Scheetz Todd E; Roos Benjamin R; Ritch Robert; West Sheila K; Kawase Kazuhide; Shire Abdirashid M; Mullins Robert F; Stone Edwin M
 CS Department of Ophthalmology and Visual Sciences, Carver College of Medicine, University of Iowa, Iowa City 52242, USA.
 SO Human mutation, (2006 Sep) Vol. 27, No. 9, pp. 921-5.
 Journal code: 9215429. E-ISSN: 1098-1004.
 CY United States
 DT (COMPARATIVE STUDY)
 Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 LA English
 FS Priority Journals
 EM 200610
 ED Entered STN: 31 Aug 2006
 Last Updated on STN: 24 Oct 2006
 Entered Medline: 24 Oct 2006

AB Age-related macular degeneration (AMD) is the most common cause of irreversible visual loss in the developed world. Previous studies have demonstrated that the c.1204T>C, p.Tyr402His allelic variant in the complement factor H (CFH) gene is associated with an approximately three-fold increased risk for AMD in Caucasians of predominantly European descent. Both the prevalence as well as the phenotypic spectrum of AMD varies widely among persons of different ethnicities. We hypothesized that populations with a lower prevalence of AMD might also have a lower prevalence of the CFH risk allele. In this study we sought to determine the frequency of this sequence variant in control populations of Caucasians, African Americans, Hispanics, Somalis, and Japanese. Normal control populations were assembled for each ethnic group: Caucasian (n=148), Somali (n=128), African American (n=75), Hispanic (n=81), and Japanese (n=82). Individuals were genotyped using a restriction digest assay and the frequency of the C allele at nucleotide position 1204 of the CFH gene was determined. A bioinformatic approach was used to **identify SNPs** in linkage disequilibrium with rs1061170 (c.1204T>C, p.Tyr402His) from the human **haplotype** map project **database** (HapMap) in order to validate the findings. We found widely discordant frequencies of the risk allele between some of the different ethnic groups: Japanese 0.07+/-0.02, Hispanics 0.17+/-0.03, African-Americans 0.35+/-0.04, Caucasians 0.34+/-0.03, and Somalis 0.34+/-0.03. Allele frequencies generated by analysis of the HapMap **database** were consistent with these findings. This study suggests that there are other yet unidentified genetic factors important in the pathogenesis of AMD that may mitigate the effects of c.1204T>C, p.Tyr402His variant.
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L3 ANSWER 9 OF 41 MEDLINE on STN DUPLICATE 6

Full Text

AN 2006648145 MEDLINE

DN PubMed ID: 16955255

TI Association study on chromosome 20q11.21-13.13 locus and its contribution to type 2 diabetes susceptibility in Japanese.

AU Tanahashi Toshihito; Osabe Dai; Nomura Kyoko; Shinohara Shuichi; Kato Hitoshi; Ichiishi Eiichiro; Nakamura Naoto; Yoshikawa Toshikazu; Takata Yoichiro; Miyamoto Tatsuro; Shiota Hiroshi; Keshavarz Parvaneh; Yamaguchi Yuka; Kunika Kiyoshi; Moritani Maki; Inoue Hiroshi; Itakura Mitsuo

CS Division of Genetic Information, Institute for Genome Research, The University of Tokushima, 3-18-15, Tokushima, Japan.

SO Human genetics, (2006 Nov) Vol. 120, No. 4, pp. 527-42. Electronic Publication: 2006-09-06.

Journal code: 7613873. ISSN: 0340-6717.

CY Germany: Germany, Federal Republic of

DT Journal; Article; (JOURNAL ARTICLE)
(RESEARCH SUPPORT, NON-U.S. GOV'T)

LA English

FS Priority Journals

EM 200704

ED Entered STN: 4 Nov 2006

Last Updated on STN: 1 May 2007

Entered Medline: 30 Apr 2007

AB Several linkage studies have predicted that human chromosome 20q is closely related to type 2 diabetes, but there is no clear evidence that certain variant(s) or gene(s) have strong effects on the **disease** within this region. To examine **disease** susceptibility variant in Japanese, verified **SNPs** from the **databases**, with a minor allele frequency larger than 0.15, were selected at 10-kb intervals across a 19.31-Mb region (20q11.21-13.13), which contained 291 genes, including hepatocyte nuclear factor 4alpha (HNF4alpha). As a result, a total of 1,147 **SNPs** were genotyped with TaqMan assay using 1,818 Japanese samples. By searching for HNF4alpha as a representative **disease**-susceptible gene, no variants of HNF4alpha were strongly associated with **disease**. To **identify** other genetic variant related with **disease**, we designed an extensive two-stage association study (725 first and 1,093 second test samples). Although SNP1146 (rs220076) was selected as a landmark within the 19.31 Mb region, the magnitude of the nominal P value (P = 0.0023) was rather weak. Subsequently, a **haplotype**-based association study showed that two common **haplotypes** were weakly associated with **disease**. All of these tests resulted in non-significance after adjusting for Bonferroni's correction and the false discovery rate to control for the impact of multiple testing. Contrary to the initial expectations, we could not conclude that

certain **SNPs** had a major effect on this promising locus within the framework presented here. As a way to extend our observations, we emphasize the importance of a subsequent association study including replication and/or meta-analysis in multiple populations.

L3 ANSWER 10 OF 41 MEDLINE on STN

Full Text

AN 2006725692 MEDLINE

DN PubMed ID: 17137522

TI iHAP--integrated **haplotype** analysis pipeline for characterizing the **haplotype** structure of genes.

AU Song Chun Meng; Yeo Boon Huat; Tantoso Erwin; Yang Yuchen; Lim Yun Ping; Li Kuo-Bin; Rajagopal Gunaretnam

CS Bioinformatics Institute, 30 Biopolis Street, #07-01, 138671, Singapore. alfreds@bii.a-star.edu.sg. <alfreds@bii.a-star.edu.sg>

SO BMC bioinformatics, (2006) Vol. 7, pp. 525. Electronic Publication: 2006-12-01.

Journal code: 100965194. E-ISSN: 1471-2105.

CY England: United Kingdom

DT Journal; Article; (JOURNAL ARTICLE)
(RESEARCH SUPPORT, NON-U.S. GOV'T)

LA English

FS Priority Journals

EM 200701

ED Entered STN: 14 Dec 2006

Last Updated on STN: 5 Jan 2007

Entered Medline: 4 Jan 2007

AB BACKGROUND: The advent of genotype data from large-scale efforts that catalog the genetic variants of different populations have given rise to new avenues for multifactorial **disease** association studies. Recent work shows that genotype data from the International HapMap Project have a high degree of transferability to the wider population. This implies that the design of genotyping studies on local populations may be facilitated through inferences drawn from information contained in HapMap populations. RESULTS: To facilitate analysis of HapMap data for characterizing the **haplotype** structure of genes or any chromosomal regions, we have developed an integrated web-based resource, iHAP. In addition to incorporating genotype and **haplotype** data from the International HapMap Project and gene information from the UCSC Genome Browser **Database**, iHAP also provides capabilities for inferring **haplotype** blocks and selecting tag **SNPs** that are representative of **haplotype** patterns. These include block partitioning algorithms, block definitions, tag **SNP** definitions, as well as **SNPs** to be "force included" as tags. Based on the parameters defined at the input stage, iHAP performs on-the-fly analysis and displays the result graphically as a webpage. To facilitate analysis, intermediate and final result files can be downloaded. CONCLUSION: The iHAP resource, available at <http://ihap.bii.a-star.edu.sg>, provides a convenient yet flexible approach for the user community to analyze HapMap data and **identify** candidate **targets** for genotyping studies.

L3 ANSWER 11 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2006:325952 CAPLUS

DN 144:410613

TI PRKCA and multiple sclerosis: association in two independent populations

AU Saarela, Janna; Kallio, Suvi P.; Chen, Daniel; Montpetit, Alexandre; Jokiahho, Anne; Choi, Eva; Asselta, Rosanna; Bronnikov, Denis; Lincoln, Matthew R.; Sadovnick, A. Dessa; Tienari, Pentti J.; Koivisto, Keijo; Palotie, Aarno; Ebers, George C.; Hudson, Thomas J.; Peltonen, Leena

CS Department of Molecular Medicine, National Public Health Institute, Helsinki, Finland

SO PLoS Genetics (2006), 2(3), 364-375

CODEN: PGLEB5; ISSN: 1553-7404

URL: <http://genetics.plosjournals.org/archive/1553-7404/2/3/pdf/10.1371.1553-7404.2.3.complete.pdf>

PB Public Library of Science

DT Journal; (online computer file)

LA English

AB Multiple sclerosis (MS) is a chronic **disease** of the central nervous system responsible for a large portion of neurol. disabilities in young adults. Similar to what occurs in numerous complex **diseases**, both unknown environmental factors and genetic predisposition are required to

generate MS. We ascertained a set of 63 Finnish MS families, originating from a high-risk region of the country, to **identify** a susceptibility gene within the previously established 3.4-Mb region on 17q24. Initial single nucleotide polymorphism (**SNP**)-based assocn. implicated PRKCA (protein kinase C alpha) gene, and this assocn. was replicated in an independent set of 148 Finnish MS families ($p = 0.0004$; remaining significant after correction for multiple testing). Further, a dense set of 211 **SNPs** evenly covering the PRKCA gene and the flanking regions was selected from the dbSNP **database** and analyzed in two large, independent MS cohorts: in 211 Finnish and 554 Canadian MS families. A multipoint **SNP** anal. indicated linkage to PRKCA and its telomeric flanking region in both populations, and **SNP haplotype** and genotype combination analyses revealed an allelic variant of PRKCA, which covers the region between introns 3 and 8, to be over-represented in Finnish MS cases (odds ratio = 1.34, 95% confidence interval 1.07-1.68). A second allelic variant, covering the same region of the PRKCA gene, showed somewhat stronger evidence for assocn. in the Canadian families (odds ratio = 1.64, 95% confidence interval 1.39-1.94). Initial functional relevance for **disease** predisposition was suggested by the expression anal.: The transcript levels of PRKCA showed correlation with the copy no. of the Finnish and Canadian "risk" **haplotypes** in CD4-neg. mononuclear cells of five Finnish multiplex families and in lymphoblast cell lines of 11 Center d'Etude du Polymorphisme Humain (CEPH) individuals of European origin.

RE.CNT 49 THERE ARE 49 CITED REFERENCES AVAILABLE FOR THIS RECORD
 ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 12 OF 41 MEDLINE on STN

Full Text

AN 2006246514 MEDLINE

DN PubMed ID: 16501185

TI An XML-based system for synthesis of data from disparate **databases**.

AU Kurc Tahsin; Janies Daniel A; Johnson Andrew D; Langella Stephen; Oster Scott; Hastings Shannon; Habib Farhat; Camerlengo Terry; Ervin David; Catalyurek Umit V; Saltz Joel H

CS Biomedical Informatics Department, Ohio State University, 3184 Graves Hall, 333 West 10th Avenue, Columbus, OH 43210, USA.. kurc@bmi.osu.edu

NC P20EB000591 (United States NIBIB)

SO Journal of the American Medical Informatics Association : JAMIA, (2006 May-Jun) Vol. 13, No. 3, pp. 289-301. Electronic Publication: 2006-02-24. Journal code: 9430800. ISSN: 1067-5027.

CY United States

DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 (RESEARCH SUPPORT, U.S. GOV'T, NON-P.H.S.)

LA English

FS Priority Journals

EM 200606

ED Entered STN: 4 May 2006

 Last Updated on STN: 3 Jun 2006

 Entered Medline: 2 Jun 2006

AB Diverse data sets have become key building blocks of translational biomedical research. Data types captured and referenced by sophisticated research studies include high throughput genomic and proteomic data, laboratory data, data from imagery, and outcome data. In this paper, the authors present the application of an XML-based data management system to support integration of data from disparate data sources and large data sets. This system facilitates management of XML schemas and on-demand creation and management of XML **databases** that conform to these schemas. They illustrate the use of this system in an application for genotype-phenotype correlation analyses. This application implements a method of phenotype-genotype correlation based on phylogenetic optimization of large data sets of mouse **SNPs** and phenotypic data. The application workflow requires the management and integration of genomic information and phenotypic data from external data repositories and from the results of phenotype-genotype correlation analyses. Our implementation supports the process of carrying out a complex workflow that includes large-scale phylogenetic tree optimizations and application of Maddison's concentrated changes test to large phylogenetic tree data sets. The data management system also allows collaborators to share data in a uniform way and supports complex queries that **target** data sets.

L3 ANSWER 13 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2006:549811 CAPLUS

DN 146:24624

TI Evidence for a Genetic Basis for Altitude-Related Illness

AU Rupert, Jim L.; Koehle, Michael S.

CS School of Human Kinetics, University of British Columbia, Vancouver, BC, Can.

SO High Altitude Medicine & Biology (2006), 7(2), 150-167

CODEN: HAMBB7; ISSN: 1527-0297

PB Mary Ann Liebert, Inc.

DT Journal; General Review

LA English

AB A review. Altitude-related illnesses are a family of interrelated pulmonary, cerebral, hematol., and cardiovascular medical conditions assocd. with the diminished oxygen availability at moderate to high altitudes. The acute forms of these debilitating and potentially fatal conditions, which include acute mountain sickness (AMS), high altitude pulmonary edema (HAPE), and high altitude cerebral edema (HACE), often develop in incompletely acclimatized lowlanders shortly after ascent, whereas, the chronic conditions, such as chronic mountain sickness (CMS) and high altitude pulmonary hypertension (HAPH), usually afflict native or long-term highlanders and may reflect a loss of adaptation. Anecdotal reports of particularly susceptible people or families are frequently cited as evidence that certain individuals have an innate susceptibility (or resistance) to developing these conditions and, in recent decades, there have been a no. of studies designed to characterize the physiol. of individuals predisposed to these conditions, as well as to **identify** the specific genetic variants that contribute to this predisposition. This paper reviews the epidemiol. evidence for a genetic component to the various forms of altitude-related illness, such as innate susceptibility, familial clustering, and patterns of population susceptibility, as well as the mol. evidence for specific genetic risk factors. While the evidence supports some role for genetic background in the etiol. of altitude-related illness, limitations in individual studies and a general lack of corroborating research limit the conclusions that can be drawn about the extent of this contribution and the specific genes or pathways involved. The paper closes with suggestions for future work that could support and expand on previous studies, as well as provide new insights.

RE.CNT 99 THERE ARE 99 CITED REFERENCES AVAILABLE FOR THIS RECORD

ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 14 OF 41 MEDLINE on STN

Full Text

AN 2007027094 MEDLINE

DN PubMed ID: 16945141

TI Fine mapping of genes within the IDDM8 region in rheumatoid arthritis.

AU Hinks Anne; Barton Anne; John Sally; Shephard Neil; Worthington Jane

CS Arthritis Research Campaign Epidemiology Unit, University of Manchester, Manchester M13 9PT, UK.. Anne.Hinks@manchester.ac.uk

SO Arthritis research & therapy, (2006) Vol. 8, No. 5, pp. R145.

Journal code: 101154438. E-ISSN: 1478-6362.

CY England: United Kingdom

DT Journal; Article; (JOURNAL ARTICLE)

(RESEARCH SUPPORT, NON-U.S. GOV'T)

LA English

FS Priority Journals

EM 200702

ED Entered STN: 17 Jan 2007

Last Updated on STN: 10 Feb 2007

Entered Medline: 9 Feb 2007

AB The IDDM8 region on chromosome 6q27, first identified as a susceptibility locus for type 1 diabetes, has previously been linked and associated with rheumatoid arthritis (RA). The region contains a number of potential candidate genes, including programmed cell death 2 (PDCD2), the proteasome subunit beta type 1 (PSMB1), delta-like ligand 1 (DLL-1) and TATA box-binding protein (TBP) amongst others. The aim of this study was to fine map the IDDM8 region on chromosome 6q27, focusing on the genes in the region, to **identify** polymorphisms that may contribute to susceptibility to RA and potentially to other autoimmune **diseases**. Validated single nucleotide polymorphisms (**SNPs**; n = 65) were selected from public **databases** from the 330 kb region of IDDM8. These were genotyped using

Sequenom MassArray genotyping technology in two datasets; the test dataset comprised 180 RA cases and 180 controls. We tested 50 **SNPs** for association with RA and any significant associations were genotyped in a second dataset of 174 RA cases and 192 controls, and the datasets were combined before analysis. Association analysis was performed by chi-square test implemented in Stata software and linkage disequilibrium and **haplotype** analysis was performed using Helix tree version 4.1. There was initial weak evidence of association, with RA, of a number of **SNPs** around the loc154449 putative gene and within the KIAA1838 gene; however, these associations were not significant in the combined dataset. Our study has failed to detect evidence of association with any of the known genes mapping to the IDDM8 locus with RA.

L3 ANSWER 15 OF 41 MEDLINE on STN

Full Text

AN 2006190966 MEDLINE

DN PubMed ID: 16596167

TI PRKCA and multiple sclerosis: association in two independent populations.

AU Saarela Janna; Kallio Suvi P; Chen Daniel; Montpetit Alexandre; Jokiaho Anne; Choi Eva; Asselta Rosanna; Bronnikov Denis; Lincoln Matthew R; Sadovnick A Dessa; Tienari Pentti J; Koivisto Keijo; Palotie Aarno; Ebers George C; Hudson Thomas J; Peltonen Leena

CS Department of Molecular Medicine, National Public Health Institute, Helsinki, Finland.. Janna.Saarela@ktl.fi

NC NS 43559 (United States NINDS)

SO PLoS genetics, (2006 Mar) Vol. 2, No. 3, pp. e42. Electronic Publication: 2006-03-31.

Journal code: 101239074. E-ISSN: 1553-7404.

CY United States

DT Journal; Article; (JOURNAL ARTICLE)
(RESEARCH SUPPORT, N.I.H., EXTRAMURAL)

LA English

FS Priority Journals

EM 200608

ED Entered STN: 6 Apr 2006

Last Updated on STN: 4 Aug 2006

Entered Medline: 3 Aug 2006

AB Multiple sclerosis (MS) is a chronic **disease** of the central nervous system responsible for a large portion of neurological disabilities in young adults. Similar to what occurs in numerous complex **diseases**, both unknown environmental factors and genetic predisposition are required to generate MS. We ascertained a set of 63 Finnish MS families, originating from a high-risk region of the country, to **identify** a susceptibility gene within the previously established 3.4-Mb region on 17q24. Initial single nucleotide polymorphism (**SNP**)-based association implicated PRKCA (protein kinase C alpha) gene, and this association was replicated in an independent set of 148 Finnish MS families ($p = 0.0004$; remaining significant after correction for multiple testing). Further, a dense set of 211 **SNPs** evenly covering the PRKCA gene and the flanking regions was selected from the dbSNP **database** and analyzed in two large, independent MS cohorts: in 211 Finnish and 554 Canadian MS families. A multipoint **SNP** analysis indicated linkage to PRKCA and its telomeric flanking region in both populations, and **SNP haplotype** and genotype combination analyses revealed an allelic variant of PRKCA, which covers the region between introns 3 and 8, to be over-represented in Finnish MS cases (odds ratio = 1.34, 95% confidence interval 1.07-1.68). A second allelic variant, covering the same region of the PRKCA gene, showed somewhat stronger evidence for association in the Canadian families (odds ratio = 1.64, 95% confidence interval 1.39-1.94). Initial functional relevance for **disease** predisposition was suggested by the expression analysis: The transcript levels of PRKCA showed correlation with the copy number of the Finnish and Canadian "risk" **haplotypes** in CD4-negative mononuclear cells of five Finnish multiplex families and in lymphoblast cell lines of 11 Centre d'Etude du Polymorphisme Humain (CEPH) individuals of European origin.

L3 ANSWER 16 OF 41 MEDLINE on STN

Full Text

AN 2006252703 MEDLINE

DN PubMed ID: 16597333

TI Selection of **SNP** subsets for association studies in candidate genes: comparison of the power of different strategies to detect single **disease**

susceptibility locus effects.

AU Cousin Emmanuelle; Deleuze Jean-Francois; Genin Emmanuelle
 CS Sanofi-Aventis, Evry Genetics Centre, 2 rue Gaston Cremieux CP5705, 91057
 Evry, France.. emmanuelle.cousin@sanofi-aventis.com
 SO BMC genetics, (2006) Vol. 7, pp. 20. Electronic Publication: 2006-04-05.
 Journal code: 100966978. E-ISSN: 1471-2156.
 CY England: United Kingdom
 DT (COMPARATIVE STUDY)
 Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Priority Journals
 EM 200605
 ED Entered STN: 9 May 2006
 Last Updated on STN: 19 May 2006
 Entered Medline: 18 May 2006

AB BACKGROUND: The recent advances in genotyping and molecular techniques have greatly increased the knowledge of the human genome structure. Millions of polymorphisms are reported and freely available in public **databases**. As a result, there is now a need to **identify** among all these data, the relevant markers for genetic association studies. Recently, several methods have been published to select subsets of markers, usually Single Nucleotide Polymorphisms (**SNPs**), that best represent genetic polymorphisms in the studied candidate gene or region. RESULTS: In this paper, we compared four of these selection methods, two based on **haplotype** information and two based on pairwise linkage disequilibrium (LD). The methods were applied to the genotype data on twenty genes with different patterns of LD and different numbers of **SNPs**. A measure of the efficiency of the different methods to select **SNPs** was obtained by comparing, for each gene and under several single **disease** susceptibility models, the power to detect an association that will be achieved with the selected **SNP** subsets. CONCLUSION: None of the four selection methods stands out systematically from the others. Methods based on pairwise LD information turn out to be the most interesting methods in a context of association study in candidate gene. In a context where the number of **SNPs** to be tested in a given region needs to be more limited, as in large-scale studies or wide genome **scans**, one of the two methods based on **haplotype** information, would be more suitable.

L3 ANSWER 17 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN
Full Text
 AN 2006:1056660 CAPLUS
 DN 146:80112
 TI Fine mapping of genes within the IDDM8 region in rheumatoid arthritis
 AU Hinks, Anne; Barton, Anne; John, Sally; Shephard, Neil; Worthington, Jane
 CS Arthritis Research Campaign Epidemiology Unit, University of Manchester, Manchester, M13 9PT, UK
 SO Arthritis Research & Therapy (2006), 8(5), No pp. given
 CODEN: ARTRCV; ISSN: 1478-6362
 URL: <http://arthritis-research.com/content/pdf/ar2037.pdf>
 PB BioMed Central Ltd.
 DT Journal; (online computer file)
 LA English

AB The IDDM8 region on chromosome 6q27, first identified as a susceptibility locus for type 1 diabetes, has previously been linked and assocd. with rheumatoid arthritis (RA). The region contains a no. of potential candidate genes, including programmed cell death 2 (PDCD2), the proteasome subunit beta type 1 (PSMB1), delta-like ligand 1 (DLL-1) and TATA box-binding protein (TBP) amongst others. The aim of this study was to fine map the IDDM8 region on chromosome 6q27, focusing on the genes in the region, to **identify** polymorphisms that may contribute to susceptibility to RA and potentially to other autoimmune **diseases**. Validated single nucleotide polymorphisms (**SNPs**; n = 65) were selected from public **databases** from the 330 kb region of IDDM8. These were genotyped using Sequenom MassArray genotyping technol. in two datasets; the test dataset comprised 180 RA cases and 180 controls. We tested 50 **SNPs** for assocn. with RA and any significant assocns. were genotyped in a second dataset of 174 RA cases and 192 controls, and the datasets were combined before anal. Assocn. anal. was performed by chi-square test implemented in Stata software and linkage disequil. and **haplotype** anal. was performed using Helix tree version 4.1. There was initial weak evidence of assocn., with RA, of a no. of **SNPs** around the loc154449 putative gene and within the KIAA1838 gene; however, these assocns. were not significant in the

combined dataset. Our study has failed to detect evidence of assocn. with any of the known genes mapping to the IDDM8 locus with RA.

RE.CNT 20 THERE ARE 20 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 18 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2006:384116 CAPLUS

DN 145:225413

TI Selection of **SNP** subsets for association studies in candidate genes: comparison of the power of different strategies to detect single **disease** susceptibility locus effects

AU Cousin, Emmanuelle; Deleuze, Jean-Francois; Genin, Emmanuelle

CS Sanofi-Aventis, Evry Genetics Centre, Evry, 91057, Fr.

SO BMC Genetics (2006), 7, No pp. given

CODEN: BGMEDS; ISSN: 1471-2156

URL: <http://www.biomedcentral.com/content/pdf/1471-2156-7-20.pdf>

PB BioMed Central Ltd.

DT Journal; (online computer file)

LA English

AB The recent advances in genotyping and mol. techniques have greatly increased the knowledge of the human genome structure. Millions of polymorphisms are reported and freely available in public **databases**. As a result, there is now a need to **identify** among all these data, the relevant markers for genetic assocn. studies. Recently, several methods have been published to select subsets of markers, usually Single Nucleotide Polymorphisms (**SNPs**), that best represent genetic polymorphisms in the studied candidate gene or region. In this paper, we compared four of these selection methods, two based on **haplotype** information and two based on pairwise linkage disequil. (LD). The methods were applied to the genotype data on twenty genes with different patterns of LD and different nos. of **SNPs**. A measure of the efficiency of the different methods to select **SNPs** was obtained by comparing, for each gene and under several single **disease** susceptibility models, the power to detect an assocn. that will be achieved with the selected **SNP** subsets. Conclusions None of the four selection methods stands out systematically from the others. Methods based on pairwise LD information turn out to be the most interesting methods in a context of assocn. study in candidate gene. In a context where the no. of **SNPs** to be tested in a given region needs to be more limited, as in large scale studies or wide genome **scans**, one of the two methods based on **haplotype** information, would be more suitable.

RE.CNT 19 THERE ARE 19 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 19 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2005:1242662 CAPLUS

DN 143:455599

TI Functional proteomics modeling system

IN Solomon, Neal

PA USA

SO U.S. Pat. Appl. Publ., 68 pp.

CODEN: USXXCO

DT Patent

LA English

FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
	-----	---	-----	-----	-----
PI	US 2005260663	A1	20051124	US 2005-133492	20050519
PRAI	US 2004-572716P	P	20040519		

AB The invention develops models of functional proteomics. Simulation scenarios of protein pathway vectors and protein-protein interactions are modeled from limited information in protein **databases**. The system focuses on three integrated subsystems, including (1) a system to model protein-protein interactions using an evolvable Global Proteomic Model (GPM) of functional proteomics to ascertain healthy pathway operations, (2) a system to **identify haplotypes** customized for specific pathol. using dysfunctional protein pathway simulations of the function of combinations of single nucleotide polymorphisms (**SNPs**) so as to ascertain pathol. mutation sources and (3) a pharmacoproteomic modeling system to develop, test and refine proposed **drug** solns. based on the

mol. structure and topol. of mutant protein(s) in order to manage individual pathologies. The system focuses on simulating the degenerative genetic **disease** categories of cancer, neurodegenerative **diseases**, immunodegenerative **diseases** and aging. The system reveals approaches to reverse engineer and test personalized medicines based upon dysfunctional proteomic pathol. simulations.

L3 ANSWER 20 OF 41 MEDLINE on STN

Full Text

AN 2005493741 MEDLINE

DN PubMed ID: 16166290

TI Five loci, SLT1 to SLT5, controlling the susceptibility to spontaneously occurring lung cancer in mice.

AU Wang Daolong; You Ming

CS Department of Surgery and the Siteman Cancer Center, Washington University School of Medicine, St. Louis, Missouri 63110, USA.

NC CA099147 (United States NCI)

CA099187 (United States NCI)

ES012063 (United States NIEHS)

ES013340 (United States NIEHS)

SO Cancer research, (2005 Sep 15) Vol. 65, No. 18, pp. 8158-65.

Journal code: 2984705R. ISSN: 0008-5472.

CY United States

DT Journal; Article; (JOURNAL ARTICLE)

(RESEARCH SUPPORT, N.I.H., EXTRAMURAL)

(RESEARCH SUPPORT, U.S. GOV'T, P.H.S.)

LA English

FS Priority Journals

EM 200511

ED Entered STN: 17 Sep 2005

Last Updated on STN: 15 Dec 2005

Entered Medline: 25 Nov 2005

AB A series of linkage studies was previously conducted to **identify** quantitative trait loci associated with chemically induced lung tumors. However, little is known of genetic susceptibility to spontaneously occurring lung tumorigenesis (SLT) in mice. In this study, we did a whole-genome linkage disequilibrium analysis for susceptibility to SLT in mice using approximately 135,900 single-nucleotide polymorphisms (**SNPs**) from the Roche and Genomic Institute of the Novartis Research Foundation **SNP databases**. A common set of 13 mouse strains was used, including 10 resistant strains (129X1/SvJ, AKR/J, C3H/HeJ, C57BL/6J, DBA/2J, NZB/BlnJ, CAST/EiJ, SPRET/EiJ, SM/J, and LP/J) and 3 susceptible strains (A/J, BALB/cJ, and NZW/LaCJ). Fisher exact test was used to assess the association between individual **SNPs** and susceptibility to SLT. Five regions, SLT1 to SLT5, were mapped on chromosomes 6, 7, 8, 19, and X, respectively. SLT1 to SLT5 showed a significant association with SLT under the empirical threshold ($P \leq 0.004$) derived from permutation tests. **SNP** versus **SNP** association tests indicated that these SLT regions were unlikely to be caused by population substructure. Thus, SLT1 to SLT5 seem to be novel loci controlling the susceptibility to spontaneously occurring lung cancer in mice. Our results provide, for the first time, an insight into the genetic control of spontaneously occurring lung tumorigenesis.

L3 ANSWER 21 OF 41 MEDLINE on STN

DUPLICATE 7

Full Text

AN 2005348501 MEDLINE

DN PubMed ID: 16000574

TI A **haplotype** analysis of HER-2 gene polymorphisms: association with breast cancer risk, HER-2 protein expression in the tumor, and **disease** recurrence in Korea.

AU Han Wonshik; Kang Daehee; Lee Jong Eun; Park In Ae; Choi Ji-Yeob; Lee Kyung-Mu; Bae Ji Yeon; Kim Sook; Shin Eun-Soon; Lee Jeong Eon; Shin Hyuk-Jae; Kim Seok Won; Kim Sung-Won; Noh Dong-Young

CS Department of Surgery, Seoul National University College of Medicine, and DNA Link Inc., Seoul, Korea.

SO Clinical cancer research : an official journal of the American Association for Cancer Research, (2005 Jul 1) Vol. 11, No. 13, pp. 4775-8.

Journal code: 9502500. ISSN: 1078-0432.

CY United States

DT Journal; Article; (JOURNAL ARTICLE)

(RESEARCH SUPPORT, NON-U.S. GOV'T)

LA English
 FS Priority Journals
 EM 200512
 ED Entered STN: 8 Jul 2005
 Last Updated on STN: 15 Dec 2005
 Entered Medline: 7 Dec 2005

AB PURPOSE: A single-nucleotide polymorphism (**SNP**) in codon 655 of HER-2 has been extensively studied with inconclusive results. The purpose of this study was to investigate the association between common variants of HER-2 and breast cancer risk, HER-2 expression, and survival using a **haplotype**-based stepwise approach. EXPERIMENTAL DESIGN: Twenty-nine **SNPs** listed in the National Center for Biotechnology Information **database** were screened to **identify** novel polymorphisms of HER-2 gene in 90 healthy Korean women. Six of 29 **SNPs** were polymorphic and had greater than 10% of minor allele frequencies. Using these six **SNPs**, linkage disequilibrium and **haplotype** patterns were characterized. We tested association between the **haplotypes** and breast cancer in a large case-control study (n=1,039 cases and 995 controls). Six-hundred two breast cancer patients with follow-up at least 24 months were analyzed for outcome in relation to **haplotype**. Expression of HER-2 protein was determined by immunohistochemistry in 1,094 cases of invasive breast cancer. RESULTS: All six **SNPs** showed a strong linkage disequilibrium pattern and were considered to belong to one **haplotype** block. Two **haplotype**-tagging **SNPs** (I655V and P1170A) for three common **haplotypes** (>5%) were genotyped in cases and controls. The **haplotypes** and individual **SNPs** were not associated with breast cancer risk. In patients with at least one copy of **haplotype** I (the most common **haplotype**), HER-2 expression was 1.5 times higher (P = 0.009) and the prognosis was worse (P = 0.032) compared with patients without having that **haplotype**. CONCLUSIONS: Our results suggest that the currently identified genetic polymorphisms of HER-2 are not associated with an increased risk of breast cancer in Korean women, whereas one **haplotype** does affect protein expression of the tumor and **disease** outcome.

L3 ANSWER 22 OF 41 MEDLINE on STN
Full Text
 AN 2005574911 MEDLINE
 DN PubMed ID: 16255080
 TI A **haplotype** map of the human genome.
 AU Anonymous
 CS International HapMap Consortium.
 NC R01 HG001720-06 (United States NHGRI)
 SO Nature, (2005 Oct 27) Vol. 437, No. 7063, pp. 1299-320.
 Journal code: 0410462. E-ISSN: 1476-4687.
 CY England: United Kingdom
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 (RESEARCH SUPPORT, U.S. GOV'T, P.H.S.)

LA English
 FS Priority Journals
 EM 200511
 ED Entered STN: 29 Oct 2005
 Last Updated on STN: 16 Dec 2005
 Entered Medline: 23 Nov 2005

AB Inherited genetic variation has a critical but as yet largely uncharacterized role in human **disease**. Here we report a public **database** of common variation in the human genome: more than one million single nucleotide polymorphisms (**SNPs**) for which accurate and complete genotypes have been obtained in 269 DNA samples from four populations, including ten 500-kilobase regions in which essentially all information about common DNA variation has been extracted. These data document the generality of recombination hotspots, a block-like structure of linkage disequilibrium and low **haplotype** diversity, leading to substantial correlations of **SNPs** with many of their neighbours. We show how the HapMap resource can guide the design and analysis of genetic association studies, shed light on structural variation and recombination, and **identify** loci that may have been subject to natural selection during human evolution.

L3 ANSWER 23 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN
Full Text

AN 2005:1148839 CAPLUS
DN 144:65084
TI A **haplotype** map of the human genome
AU Gibbs, Richard A.; Belmont, John W.; Boudreau, Andrwe; Leal, Suzanne M.; Hardenbol, Paul; Paternak, Shiran; Wheeler, David A.; Willis, Thomas D.; Yu, Fuli; Yang, Huanming; Zeng, Changqing; Gao, Yang; Hu, Haoran; Hu, Weitao; Li, Chaohua; Lin, Wei; Liu, Siqi; Pan, Hao; Tang, Xiaoli; Wang, Jian; Wang, Wei; Yu, Jun; Zhang, Bo; Zhang, Qingrun; Zhao, Hongbin; Zhao, Hui; Zhou, Jun; Gabriel, Stacey B.; Barry, Rachel; Blumenstiel, Brendan; Camargo, Amy; Defelice, Matthew; Faggart, Maura; Govette, Mary; Gupta, Supriya; Moore, Jamie; Nguyen, Huy; Onofrio, Robert C.; Parkin, Melissa; Roy, Jessica; Stahl, Erich; Winchester, Ellen; Ziaugra, Liuda; Atlshuler, David; Shen, Yan; Yao, Zhijian; Huang, Wei; Chu, Xun; He, Yungang; Jin, Li; Liu, Yangfan; Shen, Yayun; Sun, Weiwei; Wang, Haifeng; Wang, Yi; Wang, Ying; Wang, Ying; Xiong, Xiaoyan; Xu, Liang; Waye, Mary M. Y.; Tsui, Stephen K.; Xue, Hong; Wong, J. Tze-Fei; Galver, Launa M.; Fan, Jian-Bing; Murray, Sarah S.; Oliphant, Arnold R.; Chee, Mark S.; Montpetit, Alexandre; Chagnon, Fanny; Ferretti, Vincent; Leboeuf, Martin; Olivier, Jean-Francois; Phillips, Michael; Roumy, Stephanie; Sallee, Clementine; Verner, Andrei; Hudson, Thomas J.; Frazer, Kelly A.; Ballinger, Dennis G.; Cox, David R.; Hinds, David A.; Stuve, Laura L.; Kwok, Pui-Yan; Cai, Dongmei; Koboldt, Daniel C.; Miller, Raymond D.; Pawlikowska, Ludmila; Taillon-Miller, Patricia; Xiao, Ming; Tsui, Lap-Chee; Mak, William; Sham, Pak C.; Song, You Qiang; Tam, Paul K. H.; Nakamura, Yusuke; Kawaguchi, Takahisa; Kitamoto, Takuya; Morizon, Takashi; Nagashima, Atsushi; Ohnishi, Yozo; Sekine, Akihiro; Tanaka, Toshihiro; Tsunoda, Tatsuhiko; Deloukas, Panos; Bird, Christine P.; Delgado, Marcos; Dermitzakis, Emmanouil T.; Gwilliam, Rhian; Hunt, Sarah; Morrison, Jonathon; Powell, Don; Stranger, Barbara E.; Whittaker, Pamela; Bentley, David R.; Daly, Mark J.; de Bakker, Paul I. W.; Barrett, Jeff; Fry, Ben; Maller, Julian; McCarroll, Steve; Patterson, Nick; Pe'er, Itsik; Purcell, Shaun; Richter, Daniel; Sabeti, Pardis; Saxena, Richa; Schaffner, Stephen F.; Varilly, Patrick; Altshuler, David; Stein, Lincoln D.; Krishnan, Lalitha; Smith, Albert Vernon; Thorisson, Gudmundur A.; Chakravarti, Aravinda; Chen, Peter E.; Cutler, David J.; Kashuk, Carl S.; Lin, Shin; Abecasis, Goncalo R.; Guan, Weihua; Munro, Heather M.; Qin, Zhaohui Steve; Thomas, Daryl J.; McVean, Gilean; Bottolo, Leonardo; Eyheramendy, Susana; Freeman, Colin; Marchini, Jonathan; Myers, Simon; Spencer, Chris; Spehens, Matthew; Donnelly, Peter; Cardon, Lon R.; Clarke, Geraldine; Evans, David M.; Morris, Andrew P.; Weir, Bruce S.; Tsunoda, Tatsuhiko; Mullikin, James C.; Sherry, Stephen T.; Feolo, Michael; Zahng, Houcan; Zeng, Changqing; Zhao, Hui; Matsuda, Ichiro; Fukushima, Yoshimitsu; Macer, Darryl R.; Suda, Eiko; Rotimi, Charles N.; Adebamowo, Clement A.; Ajayi, Ike; Aniagwu, Toyin; Marshall, Patricia A.; Nkwdimmah, Chibuzor; Royal, Charmaine D.; Leppert, Mark F.; Dixon, Missy; Peiffer, Andy; Qui, Renzong; Kent, Alastair; Kato, Kazuto; Niikawa, Norio; Adewole, Isaac F.; Knoppers, Bartha M.; Foster, Morris W.; Clayton, Ellen Wright; Watkin, Jessica; Muzny, Donna; Nazareth, Lynne; Sodergren, Erica; Weinstock, George M.; Wheeler, David M.; Yakub, Imtiaz; Gabriel, Stacey B.; Onofrio, Robert C.; Richter, Daniel J.; Ziaugra, Liuda; Birren, Bruce W.; Wilson, Richard K.; Fulton, Lucinda L.; Rogers, Jane; Burton, John; Carter, Nigel P.; Clee, Christopher M.; Griffiths, Mark; Jones, Matthew C.; McLay, Kirsten; Plumb, Robert W.; Ross, Mark T.; Sims, Sarah K.; Willey, David L.; Chen, Zhu; Han, Hua; Kang, Le; Godbout, Martin; Wallenburg, John C.; Archeveque, Paul L.; Bellemare, Guy; Saeki, Koji; Wang, Hongguang; An, Daochang; Fu, Hongbo; Li, Qing; Wang, Zhen; Wang, Renwu; Holden, Arthur L.; Brooks, Lisa D.; McEwen, Jean E.; Bird, Christianne R.; Guyer, Mark S.; Nailer, Patrick J.; Wang, Vivian Ota; Peterson, Jane L.; Shi, Michael; Spiegel, Jack; Sung, Lawrence M.; Witonsky, Jonathan; Zacharia, Lynn F.; Collins, Francis S.; Kennedy, Karen; Jamieson, Ruth; Stewart, John

CS International HapMap Consortium, Human Genome Sequencing Center, Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, 77030, USA

SO Nature (London, United Kingdom) (2005), 437(7063), 1299-1320
CODEN: NATUAS; ISSN: 0028-0836

PB Nature Publishing Group
DT Journal
LA English
AB Inherited genetic variation has a crit. but as yet largely uncharacterized role in human **disease**. Here we report a public **database** of common variation in the human genome: more than one million single nucleotide polymorphisms (**SNPs**) for which accurate and complete genotypes have been

obtained in 269 DNA samples from four populations, including ten 500-kilobase regions in which essentially all information about common DNA variation has been extd. These data document the generality of recombination hotspots, a block-like structure of linkage disequil. and low **haplotype** diversity, leading to substantial correlations of **SNPs** with many of their neighbors. We show how the HapMap resource can guide the design and anal. of genetic assocn. studies, shed light on structural variation and recombination, and **identify** loci that may have been subject to natural selection during human evolution.

RE.CNT 103 THERE ARE 103 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 24 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2005:1284119 CAPLUS

DN 144:32710

TI Genome sequence, comparative analysis and **haplotype** structure of the domestic dog

AU Lindblad-Toh, Kerstin; Wade, Claire M.; Mikkelsen, Tarjei S.; Karlsson, Elinor K.; Jaffe, David B.; Kamal, Michael; Clamp, Michele; Chang, Jean L.; Kulbokas, Edward J.; Zody, Michael C.; Mauceli, Evan; Xie, Xiaohui; Breen, Matthew; Wayne, Robert K.; Ostrander, Elaine A.; Ponting, Chris P.; Galibert, Francis; Smith, Douglas R.; de Jong, Pieter J.; Kirkness, Ewen; Alvarez, Pablo; Biagi, Tara; Brockman, William; Butler, Jonathan; Chin, Chee-Wye; Cook, April; Cuff, James; Daly, Mark J.; DeCaprio, David; Gnerre, Sante; Grabherr, Manfred; Kellis, Manolis; Kleber, Michael; Bardeleben, Carolyne; Goodstadt, Leo; Heger, Andreas; Hitte, Christophe; Kim, Lisa; Koepfli, Klaus-Peter; Parker, Heidi G.; Pollinger, John P.; Searle, Stephen M. J.; Sutter, Nathan B.; Thomas, Rachael; Webber, Caleb; Baldwin, Jennifer; Abebe, Adal; Abouelleil, Amr; Aftuck, Lynne; Ait-zahra, Mostafa; Aldredge, Tyler; Allen, Nicole; An, Peter; Anderson, Scott; Antoine, Claudel; Arachchi, Harindra; Aslam, Ali; Ayotte, Laura; Bachantsang, Pasang; Barry, Andrew; Bayul, Tashi; Benamara, Mostafa; Berlin, Aaron; Bessette, Daniel; Blitshteyn, Berta; Bloom, Toby; Blye, Jason; Boguslavskiy, Leonid; Bonnet, Claude; Boukhgalter, Boris; Brown, Adam; Cahill, Patrick; Calixte, Nadia; Camarata, Jody; Cheshatsang, Yama; Chu, Jeffrey; Citroen, Mieke; Collymore, Alville; Cooke, Patrick; Dawoe, Tenzin; Daza, Riza; Decktor, Karin; DeGray, Stuart; Dhargay, Norbu; Dooley, Kimberly; Dooley, Kathleen; Dorje, Passang; Dorjee, Kunsang; Dorris, Lester; Duffey, Noah; Dupes, Alan; Egbiremolen, Osebhajajeme; Elong, Richard; Falk, Jill; Farina, Abderrahim; Faro, Susan; Ferguson, Diallo; Ferreira, Patricia; Fisher, Sheila; FitzGerald, Mike; Foley, Karen; Foley, Chelsea; Franke, Alicia; Friedrich, Dennis; Gage, Diane; Garber, Manuel; Gearin, Gary; Giannoukos, Georgia; Goode, Tina; Goyette, Audra; Graham, Joseph; Grandbois, Edward; Gyaltsen, Kunsang; Hafez, Nabil; Hagopian, Daniel; Hagos, Birhane; Hall, Jennifer; Healy, Claire; Hegarty, Ryan; Honan, Tracey; Horn, Andrea; Houde, Nathan; Hughes, Leanne; Hunnicutt, Leigh; Husby, M.; Jester, Benjamin; Jones, Charlien; Kamat, Asha; Kanga, Ben; Kells, Cristyn; Khazanovich, Dmitry; Kieu, Alix Chinh; Kisner, Peter; Kumar, Mayank; Lance, Krista; Landers, Thomas; Lara, Marcia; Lee, William; Leger, Jean-Pierre; Lennon, Niall; Leuper, Lisa; LeVine, Sarah; Liu, Jinlei; Liu, Xiaohong; Lokyitsang, Yeshi; Lokyitsang, Tashi; Lui, Annie; Macdonald, Jan; Major, John; Marabella, Richard; Maru, Kebede; Matthews, Charles; McDonough, Susan; Mehta, Teena; Meldrim, James; Melnikov, Alexandre; Meneus, Louis; Mihalev, Atanas; Mihova, Tanya; Miller, Karen; Mittelman, Rachel; Mlenga, Valentine; Mulrain, Leonidas; Munson, Glen; Navidi, Adam; Naylor, Jerome; Nguyen, Tuyen; Nguyen, Nga; Nguyen, Cindy; Nguyen, Thu; Nicol, Robert; Norbu, Nyima; Norbu, Choe; Novod, Nathaniel; Nyima, Tenchoe; Olandt, Peter; O'Neill, Barry; O'Neill, Keith; Osman, Sahal; Oyono, Lucien; Patti, Christopher; Perrin, Danielle; Phunkhang, Pema; Pierre, Fritz; Priest, Margaret; Rachupka, Anthony; Raghuraman, Sujaa; Rameau, Rayale; Ray, Verneda; Raymond, Christina; Rege, Filip; Rise, Cecil; Rogers, Julie; Rogov, Peter; Sahalie, Julie; Settipalli, Sampath; Sharpe, Theodore; Shea, Terrance; Sheehan, Mechele; Sherpa, Ngawang; Shi, Jianying; Shih, Diana; Sloan, Jessie; Smith, Cheryllyn; Sparrow, Todd; Stalker, John; Stange-Thomann, Nicole; Stavropoulos, Sharon; Stone, Catherine; Stone, Sabrina; Sykes, Sean; Tchuinga, Pierre; Tenzing, Pema; Tesfaye, Senait; Thoulutsang, Dawa; Thoulutsang, Yama; Topham, Kerri; Topping, Ira; Tsamla, Tsamla; Vassiliev, Helen; Venkataraman, Vijay; Vo, Andy; Wangchuk, Tsering; Wangdi, Tsering; Weiland, Michael; Wilkinson, Jane; Wilson, Adam; Yadav, Shailendra; Yang, Shuli; Yang, Xiaoping; Young, Geneva; Yu, Qing; Zainoun, Joanne; Zembek,

Lisa; Zimmer, Andrew; Lander, Eric S.
 CS Broad Institute of Harvard and MIT, Cambridge, MA, 02141, USA
 SO Nature (London, United Kingdom) (2005), 438(7069), 803-819
 CODEN: NATUAS; ISSN: 0028-0836
 PB Nature Publishing Group
 DT Journal
 LA English
 AB A high-quality draft genome sequence of the domestic dog (*Canis familiaris*) is reported, together with a dense map of single nucleotide polymorphisms (**SNPs**) across breeds. The dog is of particular interest because it provides important evolutionary information and because existing breeds show great phenotypic diversity for morphol., physiol., and behavioral traits. Sequence comparison with the primate and rodent lineages is used to shed light on the structure and evolution of genomes and genes. Notably, the majority of the most highly conserved non-coding sequences in mammalian genomes are clustered near a small subset of genes with important roles in development. Anal. of **SNPs** reveals long-range **haplotypes** across the entire dog genome, and defines the nature of genetic diversity within and across breeds. The current **SNP** map now makes it possible for genome-wide assocn. studies to **identify** genes responsible for **diseases** and traits, with important consequences for human and companion animal health. The draft genome sequence is deposited in the NCBI genome projects **database** under accession codes AAEX01000000 (CanFam1.0) and AAEX02000000 (CanFam2.0), and **SNPs** are deposited in the dbSNP **database**.
 RE.CNT 119 THERE ARE 119 CITED REFERENCES AVAILABLE FOR THIS RECORD
 ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 25 OF 41 MEDLINE on STN DUPLICATE 8
Full Text
 AN 2005540973 MEDLINE
 DN PubMed ID: 15993562
 TI Allelic variation in gene expression identified through computational analysis of the dbEST **database**.
 AU Lin Wei; Yang Howard H; Lee Maxwell P
 CS Laboratory of Population Genetics, National Cancer Institute, Bethesda, MD 20892, USA.
 SO Genomics, (2005 Nov) Vol. 86, No. 5, pp. 518-27. Electronic Publication: 2005-07-01.
 Journal code: 8800135. ISSN: 0888-7543.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Priority Journals
 EM 200512
 ED Entered STN: 12 Oct 2005
 Last Updated on STN: 30 Dec 2005
 Entered Medline: 29 Dec 2005
 AB Differential expression between the two alleles of an individual and between people with different genotypes has been commonly observed. Quantitative differences in gene expression between people may provide the genetic basis for the phenotypic difference between individuals and may be the primary cause of complex **diseases**. In this paper, we developed a computational method to **identify** genes that displayed allelic variation in gene expression in human EST libraries. To model allele-specific gene expression, we first identified EST libraries in which both A and B alleles were expressed and then identified allelic variation in gene expression based on the EST counts for each allele using a binomial test. Among 1107 **SNPs** that had a sufficient number of ESTs for the analysis, 524 (47%) displayed allelic variation in at least one cDNA library. We verified experimentally the allelic variation in gene expression for 6 of these **SNPs**. The frequency of allelic variation observed in EST libraries was similar to the previous studies using the **SNP** chip and primer extension method. We found that genes that displayed allelic variation were distributed throughout the human genome and were enriched in certain chromosome regions. The **SNPs** and genes identified in this study will provide a rich source for evaluating the effects of those **SNPs** and associated **haplotypes** in human health and **diseases**.

L3 ANSWER 26 OF 41 MEDLINE on STN
Full Text
 AN 2006007242 MEDLINE

DN PubMed ID: 16336665
 TI **SNP-VISTA**: an interactive **SNP** visualization tool.
 AU Shah Nameeta; Teplitzsky Michael V; Minovitsky Simon; Pennacchio Len A; Hugenholtz Philip; Hamann Bernd; Dubchak Inna L
 CS Institute for Data Analysis and Visualization, (IDAV), Department of Computer Science, University of California, Davis, One Shields Ave., Davis, CA 95616, USA.. nvshah@ucdavis.edu
 SO BMC bioinformatics, (2005) Vol. 6, pp. 292. Electronic Publication: 2005-12-08.
 Journal code: 100965194. E-ISSN: 1471-2105.
 CY England: United Kingdom
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 (RESEARCH SUPPORT, U.S. GOV'T, NON-P.H.S.)
 LA English
 FS Priority Journals
 EM 200601
 ED Entered STN: 6 Jan 2006
 Last Updated on STN: 27 Jan 2006
 Entered Medline: 26 Jan 2006
 AB BACKGROUND: Recent advances in sequencing technologies promise to provide a better understanding of the genetics of human **disease** as well as the evolution of microbial populations. Single Nucleotide Polymorphisms (**SNPs**) are established genetic markers that aid in the identification of loci affecting quantitative traits and/or **disease** in a wide variety of eukaryotic species. With today's technological capabilities, it has become possible to re-sequence a large set of appropriate candidate genes in individuals with a given **disease** in an attempt to **identify** causative mutations. In addition, **SNPs** have been used extensively in efforts to study the evolution of microbial populations, and the recent application of random shotgun sequencing to environmental samples enables more extensive **SNP** analysis of co-occurring and co-evolving microbial populations. The program is available at <http://genome.lbl.gov/vista/snpv.html>. RESULTS: We have developed and present two modifications of an interactive visualization tool, **SNP-VISTA**, to aid in the analyses of the following types of data: A. Large-scale re-sequence data of **disease**-related genes for discovery of associated and/or causative alleles (GeneSNP-VISTA). B. Massive amounts of ecogenomics data for studying homologous recombination in microbial populations (EcoSNP-VISTA). The main features and capabilities of **SNP-VISTA** are: 1) mapping of **SNPs** to gene structure; 2) classification of **SNPs**, based on their location in the gene, frequency of occurrence in samples and allele composition; 3) clustering, based on user-defined subsets of **SNPs**, highlighting **haplotypes** as well as recombinant sequences; 4) integration of protein evolutionary conservation visualization; and 5) display of automatically calculated recombination points that are user-editable. CONCLUSION: The main strength of **SNP-VISTA** is its graphical interface and use of visual representations, which support interactive exploration and hence better understanding of large-scale **SNP** data by the user.

L3 ANSWER 27 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2005:1111051 CAPLUS

DN 144:247821

TI The patterns of natural variation in human genes

AU Crawford, Dana C.; Akey, Dayna T.; Nickerson, Deborah A.

CS Department of Genome Sciences, University of Washington, Seattle, WA, 98195, USA

SO Annual Review of Genomics and Human Genetics (2005), 6, 287-312
 CODEN: ARGHC4; ISSN: 1527-8204

PB Annual Reviews Inc.

DT Journal; General Review

LA English

AB A review. Currently, more than 10 million DNA sequence variations have been uncovered in the human genome. The most detailed variation discovery efforts have focused on candidate genes involved in cardiovascular **disease** or in susceptibilities assocd. with exposure to environmental agents. Here we provide an overview of natural genetic variation from the literature and in 510 human candidate genes resequenced for variation discovery. The av. human gene contains 126 biallelic polymorphisms, 46 of which are common ($\geq 5\%$ minor allele frequency) and 5 of which are

found in coding regions. Using this complete picture of genetic diversity, we explore conservation, signatures of selection, and historical recombination to mine information useful for candidate gene assocn. studies. In general, we find that the patterns of human gene variation suggest that no one approach will be appropriate for genetic assocn. studies across all genes. Therefore, many different approaches may be required to **identify** the elusive genotypes assocd. with common human phenotypes.

RE.CNT 119 THERE ARE 119 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 28 OF 41 MEDLINE on STN

Full Text

AN 2005220359 MEDLINE
DN PubMed ID: 15703761
TI Polymorphisms in the interleukin 17F gene (IL17F) and asthma.
AU Ramsey C D; Lazarus R; Camargo C A Jr; Weiss S T; Celedon J C
CS Channing Laboratory, Department of Medicine, Brigham and Women's Hospital, Harvard Medical School, Boston, MA 02115, USA.
NC 5 T32 HL07427 (United States NHLBI)
K01 HL04370-01A1 (United States NHLBI)
U01 HL66795 (United States NHLBI)
SO Genes and immunity, (2005 May) Vol. 6, No. 3, pp. 236-41.
Journal code: 100953417. ISSN: 1466-4879.
CY England: United Kingdom
DT Journal; Article; (JOURNAL ARTICLE)
(RESEARCH SUPPORT, N.I.H., EXTRAMURAL)
(RESEARCH SUPPORT, NON-U.S. GOV'T)
(RESEARCH SUPPORT, U.S. GOV'T, P.H.S.)
LA English
FS Priority Journals
EM 200507
ED Entered STN: 29 Apr 2005
Last Updated on STN: 29 Jul 2005
Entered Medline: 28 Jul 2005
AB Interleukin17F (IL17F) is a regulatory cytokine for T-cell-mediated immune responses. The gene coding for IL17F (IL17F) is located on chromosome 6p, a genomic region linked to asthma and asthma-related phenotypes in multiple genome **scans**. IL17F is expressed in lung tissue, in bronchoalveolar lavage fluid from asthmatic subjects, and in activated CD4+ cells. We were thus interested in testing for association between single-nucleotide polymorphisms (**SNPs**) and **haplotypes** in IL17F and asthma. To characterize polymorphisms in IL17F, we sequenced this gene in a group of African Americans and a group of European Americans. A total of 50 **SNPs** (30 not previously reported in a public **database** (dbSNP build 118)) and two insertions/deletions were detected in IL17F; five of these polymorphisms were genotyped in participants of the Nurses' Health Study. We then tested for association between **SNPs** and **haplotypes** in IL17F and physician-diagnosed asthma in subjects with (cases) and without (control subjects) physician-diagnosed asthma. None of the **SNPs** or **haplotypes** tested in IL17F were associated with asthma. The polymorphisms identified in this study may be used in future studies of association between IL17F and phenotypes related to immune responses.

L3 ANSWER 29 OF 41 MEDLINE on STN

DUPLICATE 9

Full Text

AN 2005195631 MEDLINE
DN PubMed ID: 15780967
TI Human genome-wide screen of **haplotype**-like blocks of reduced diversity.
AU Costas Javier; Salas Antonio; Phillips Christopher; Carracedo Angel
CS Centro Nacional de Genotipado, Fundacion Publica Galega de Medicina Xenomica, Hospital Clinico Universitario, Universidade de Santiago de Compostela, E15706, Santiago de Compostela, Spain.. bfcostas@usc.es
SO Gene, (2005 Apr 11) Vol. 349, pp. 219-25.
Journal code: 7706761. ISSN: 0378-1119.
CY Netherlands
DT (COMPARATIVE STUDY)
Journal; Article; (JOURNAL ARTICLE)
(RESEARCH SUPPORT, NON-U.S. GOV'T)
LA English
FS Priority Journals
EM 200505

ED Entered STN: 15 Apr 2005
 Last Updated on STN: 28 May 2005
 Entered Medline: 27 May 2005

AB An important proportion of the human genome is organized in regions of high linkage disequilibrium (LD) and low **haplotype** diversity, referred to as **haplotype** blocks. Here, we perform a genome-wide screen of **haplotype**-like blocks presenting just two main **haplotypes** at a frequency higher than 1%, based on single-nucleotide polymorphism (SNP) frequencies from two populations: African-Americans and Caucasians, using data from the Celera **SNP database**. These **haplotype**-like blocks of reduced diversity are more abundant and of longer size in Caucasians, in agreement with population history. Several of the discovered blocks are good candidates for **targets** of natural selection, such as those blocks containing a cluster of bitter taste receptors or the apolipoprotein L1. In addition, several genes putatively involved in susceptibility to common **diseases** are included in these **haplotype**-like blocks of reduced diversity. This fact may present important implications in association studies, leading to a reduction of genotyping efforts.

L3 ANSWER 30 OF 41 MEDLINE on STN DUPLICATE 10

Full Text

AN 2005266878 MEDLINE
 DN PubMed ID: 15809674
 TI Systematic investigation of genetic variability in 111 human genes-implications for studying variable **drug** response.
 AU Freudenberg-Hua Y; Freudenberg J; Winantea J; Kluck N; Cichon S; Bruss M; Propping P; Nothen M M
 CS Institute of Human Genetics, University of Bonn, Bonn, Germany.
 SO The pharmacogenomics journal, (2005) Vol. 5, No. 3, pp. 183-92.
 Journal code: 101083949. ISSN: 1470-269X.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 LA English
 FS Priority Journals
 EM 200508
 ED Entered STN: 24 May 2005
 Last Updated on STN: 15 Aug 2005
 Entered Medline: 12 Aug 2005

AB In order to **identify** single-nucleotide polymorphisms (**SNPs**) and analyze their characteristics in a set of 111 genes, we resequenced exons and flanking regions in an average of 170 chromosomes from individuals of European origin. Genetic variability was decreased in noncoding regions highly conserved between human and rodents, indicating functional relevance of these regions. Furthermore, diversity of coding nonsynonymous **SNPs** was found lower in regions encoding a known protein sequence motif. **SNPs** predicted to be of functional significance were more common amongst rare variants. Despite the significant recent growth of **SNP** numbers in public **SNP databases**, only a small fraction of these rare variants is represented. This may be relevant in the investigation of the genetic causes of severe side effects, for which rare variants are plausible candidates. Estimation of htSNPs reduces the genotyping effort required in capturing common **haplotypes**, for certain genes, however, this accounts for only a small fraction of **haplotype** diversity.

L3 ANSWER 31 OF 41 MEDLINE on STN

Full Text

AN 2005431710 MEDLINE
 DN PubMed ID: 16029503
 TI Common variation in EMSY and risk of breast and ovarian cancer: a case-control study using HapMap tagging **SNPs**.
 AU Benusiglio Patrick R; Lesueur Fabienne; Luccarini Craig; McIntosh Joan; Luben Robert N; Smith Paula; Dunning Alison; Easton Douglas F; Ponder Bruce A J; Pharoah Paul D
 CS Strangeways Research Laboratory, Department of Oncology, University of Cambridge, Worts Causeway, Cambridge CB1 8RN, UK.. pbenusiglio@yahoo.com
 SO BMC cancer, (2005) Vol. 5, pp. 81. Electronic Publication: 2005-07-19.
 Journal code: 100967800. E-ISSN: 1471-2407.
 CY England: United Kingdom
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)

LA English
 FS Priority Journals
 EM 200602
 ED Entered STN: 16 Aug 2005
 Last Updated on STN: 28 Feb 2006
 Entered Medline: 27 Feb 2006

AB BACKGROUND: EMSY could be involved in low-level susceptibility to breast and ovarian cancer. Gene amplification is seen in a proportion of breast and ovarian tumours and correlates with poor prognosis in breast cancer patients. Furthermore, the EMSY protein silences a transcription activation domain in BRCA2 exon 3. METHODS: We used a genetic association study design to determine if common genetic variation (frequency > or = 5%) in EMSY was associated with breast or ovarian cancer risk in the British population. **Haplotype** tagging single-nucleotide polymorphisms (htSNPs) were selected from the HapMap **database** and genotyped using Taqman in two large study sets of white British women (n [breast set] = 2343 cases and 2284 controls, n [ovarian set] = 864 cases and 864 controls). HapMap data might be insufficient to tag genetic variation in EMSY comprehensively. We therefore screened the gene promoter and coding sequences with denaturing high performance liquid chromatography in order to **identify** additional **SNPs** that are most likely to be functional. RESULTS: HapMap data on 22 **SNPs** show that 4 htSNPs tag 4 common **haplotypes**: rs2282611 (5'up t > g), rs4245443 (IVS7 g > a), rs2513511 (IVS16 a > g), rs2155220 (3'down c > t). We observed no association between any of the genotypes or associated **haplotypes** and breast or ovarian cancer risk. Seventeen out of the 18 remaining HapMap polymorphisms (94%) were well tagged by the 4 selected htSNPs (r2s > 0.8). Genotype frequencies for two further **SNPs** identified by screening and located near exon-intron boundaries, rs2508740 (IVS9 a > g) and rs11600501 (IVS10 c > t), were also similar in cases and controls. In order to simulate unidentified **SNPs**, we performed the leave-one-out cross-validation procedure on the HapMap data; over 95% of the common genetic variation was well represented by tagging polymorphisms. We are therefore likely to have tagged any common, functional variants present in our population. CONCLUSION: We found no association between common genetic variation in EMSY and risk of breast or ovarian cancer in two large study sets of white British women.

L3 ANSWER 32 OF 41 MEDLINE on STN DUPLICATE 11
Full Text
 AN 2004637526 MEDLINE
 DN PubMed ID: 15580557
 TI Variations in human HM74 (GPR109B) and HM74A (GPR109A) niacin receptors.
 AU Zellner Christian; Pullinger Clive R; Aouizerat Bradley E; Frost Philip H; Kwok Pui-Yan; Malloy Mary J; Kane John P
 CS Cardiovascular Research Institute, University of California, San Francisco 94143-0130, USA.. zellner@medicine.ucsf.edu
 SO Human mutation, (2005 Jan) Vol. 25, No. 1, pp. 18-21.
 Journal code: 9215429. E-ISSN: 1098-1004.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 LA English
 FS Priority Journals
 EM 200602
 ED Entered STN: 23 Dec 2004
 Last Updated on STN: 14 Dec 2005
 Entered Medline: 27 Feb 2006

AB HM74 (GPR109B) and the highly homologous gene, HM74A (GPR109A) code for Gi-G protein-coupled orphan receptors that recently have been discovered to be involved in the metabolic effects of niacin. The B vitamin niacin is an important agent used in the treatment of dyslipidemias, but its use is limited by side effects. The novel role of the adjacent HM74 and HM74A genes in the metabolism of niacin may provide new **targets** for **drug** development. Human genetic variations in HM74 and HM74A have been reported but have not been studied in detail. These variations may play a role in the response to agents targeting receptors coded by these genes. Here we show that many of the nonsynonymous **SNPs** listed in public **databases** for HM74 and HM74A are artifacts resulting from extensive homology between these two genes. This may be representative of a neglected phenomenon in reporting sequences of highly homologous genes. We provide primer sequences that permit selective amplification of the

complete coding regions of HM74 and HM74A. Using these primers, we show that subsequent sequencing of HM74 and HM74A reveals a novel and unique variation in the HM74A gene. **Haplotype** analysis suggests four **SNPs** can define the five major **haplotypes** that lie within a single **haplotype** block encompassing these two genes.

L3 ANSWER 33 OF 41 MEDLINE on STN

Full Text

AN 2005197555 MEDLINE

DN PubMed ID: 15799788

TI Polymorphism screening and **haplotype** analysis of the tryptophan hydroxylase gene (TPH1) and association with bipolar affective disorder in Taiwan.

AU Lai Te-Jen; Wu Chia-Yen; Tsai Hsu-Wen; Lin Yi-Mei J; Sun H Sunny

CS Institute of Medicine, Chung Shan Medical University, Taichung, Taiwan, Republic of China. tejenlai@hotmail.com. <tejenlai@hotmail.com>

SO BMC medical genetics, (2005 Mar 31) Vol. 6, pp. 14. Electronic Publication: 2005-03-31.

Journal code: 100968552. E-ISSN: 1471-2350.

CY England: United Kingdom

DT Journal; Article; (JOURNAL ARTICLE)

LA English

FS Priority Journals

EM 200505

ED Entered STN: 17 Apr 2005

Last Updated on STN: 3 May 2005

Entered Medline: 2 May 2005

AB BACKGROUND: Disturbances in serotonin neurotransmission are implicated in the etiology of many psychiatric disorders, including bipolar affective disorder (BPD). The tryptophan hydroxylase gene (TPH), which codes for the enzyme catalyzing the rate-limiting step in serotonin biosynthetic pathway, is one of the leading candidate genes for psychiatric and behavioral disorders. In a preliminary study, we found that TPH1 intron7 A218C polymorphism was associated with BPD. This study was designed to investigate sequence variants of the TPH1 gene in Taiwanese and to test whether the TPH1 gene is a susceptibility factor for the BPD. METHODS: Using a systematic approach, we have searched the exons and promoter region of the TPH1 gene for sequence variants in Taiwanese Han and have identified five variants, A-1067G, G-347T, T3804A, C27224T, and A27237G. These five variants plus another five taken from the literature and a public **database** were examined for an association in 108 BPD patients and 103 controls; no association was detected for any of the 10 variants. RESULTS: **Haplotype** constructions using these 10 **SNPs** showed that the 3 most common **haplotypes** in both patients and controls were identical. One of the fourth common **haplotype** in the patient group (i.e. GGGAGACCCA) was unique and showed a trend of significance with the **disease** ($P = 0.028$). However, the significance was abolished after Bonferroni correction thus suggesting the association is weak. In addition, three **haplotype**-tagged **SNPs** (htSNPs) were selected to represent all **haplotypes** with frequencies larger than 2% in the Taiwanese Han population. The defined TPH1 htSNPs significantly reduce the marker number for **haplotype** analysis thus provides useful information for future association studies in our population. CONCLUSION: Results of this study did not support the role of TPH1 gene in BPD etiology. As the current studies found the TPH1 gene under investigation belongs to the peripheral serotonin system and may link to a cardiac dysfunction phenotype, a second TPH gene that functions predominantly in the brain (i.e., nTPH or TPH2) should be the **target** for the future association study.

L3 ANSWER 34 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2005:537962 CAPLUS

DN 143:227194

TI Polymorphism screening and **haplotype** analysis of the tryptophan hydroxylase gene (TPHI) and association with bipolar affective disorder in Taiwan

AU Lai, Te-Jen; Wu, Chia-Yen; Tsai, Hsu-Wen; Lin, Yi-Mei J.; Sun, H. Sunny

CS Institute of Medicine, Chung Shan Medical University, Taichung, Taiwan

SO BMC Medical Genetics (2005), 6, No pp. given

CODEN: BMGMAR; ISSN: 1471-2350

URL: <http://www.biomedcentral.com/content/pdf/1471-2350-6-14.pdf>

PB BioMed Central Ltd.
 DT Journal; (online computer file)
 LA English
 AB Background: Disturbances in serotonin neurotransmission are implicated in the etiol. of many psychiatric disorders, including bipolar affective disorder (BPD). The tryptophan hydroxylase gene (TPH), which codes for the enzyme catalyzing the rate-limiting step in serotonin biosynthetic pathway, is one of the leading candidate genes for psychiatric and behavioral disorders. In a preliminary study, we found that TPH1 intron 7 A218C polymorphism was assocd. with BPD. This study was designed to investigate sequence variants of the TPH1 gene in Taiwanese and to test whether the TPH1 gene is a susceptibility factor for the BPD. Methods: Using a systematic approach, we have searched the exons and promoter region of the TPH1 gene for sequence variants in Taiwanese Han and have identified five variants, A-1067G, G-347T, T3804A, C27224T, and A27237G. These five variants plus another five taken from the literature and a public **database** were examd. for an assocn. in 108 BPD patients and 103 controls; no assocn. was detected for any of the 10 variants. Results: **Haplotype** constructions using these 10 **SNPs** showed that the 3 most common **haplotypes** in both patients and controls were identical. One of the fourth common **haplotype** in the patient group (i.e. GGGAGACCCA) was unique and showed a trend of significance with the **disease** (P = 0.028). However, the significance was abolished after Bonferroni correction thus suggesting the assocn. is weak. In addn., three **haplotype**-tagged **SNPs** (htSNPs) were selected to represent all **haplotypes** with frequencies larger than 2% in the Taiwanese Han population. The defined TPH1 htSNPs significantly reduce the marker no. for **haplotype** anal. thus provides useful information for future assocn. studies in our population. Conclusion: Results of this study did not support the role of TPH1 gene in BPD etiol. As the current studies found the TPH1 gene under investigation belongs to the peripheral serotonin system and may link to a cardiac dysfunction phenotype, a second TPH gene that functions predominantly in the brain (i.e., nTPH or TPH2) should be the **target** for the future assocn. study.

RE.CNT 52 THERE ARE 52 CITED REFERENCES AVAILABLE FOR THIS RECORD
 ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 35 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2004:183046 CAPLUS
 DN 140:230610
 TI Detection of **SNPs** and **haplotype** blocks within human chromosome 21 and association with Long QT Syndrome and hearing disorders
 IN Cox, David R.; Arnold, Deana A.
 PA Perlegen Sciences, Inc., USA
 SO PCT Int. Appl., 128 pp.
 CODEN: PIXXD2
 DT Patent
 LA English
 FAN.CNT 5

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	WO 2004018701	A2	20040304	WO 2003-US26469	20030822
	WO 2004018701	A3	20050317		
	W:	AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UZ, VC, VN, YU, ZA, ZM, ZW			
	RW:	GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM, AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PT, RO, SE, SI, SK, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG			
	US 2003077633	A1	20030424	US 2002-227195	20020822
	US 7115726	B2	20061003		
	US 2006234216	A9	20061019		
	US 2004053232	A1	20040318	US 2002-227152	20020822
	WO 2004070061	A1	20040819	WO 2003-US14799	20030508
	W:	AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR,			

LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, NZ, OM, PH, PL, PT,
 RO, RU, SC, SD, SE, SG, SK, SL, TJ, TM, TN, TR, TT, TZ, UA, UG,
 US, UZ, VC, VN, YU, ZA, ZM, ZW
 RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW, AM, AZ, BY,
 KG, KZ, MD, RU, TJ, TM, AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES,
 FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PT, RO, SE, SI, SK, TR,
 BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG

AU	2003303869	A1	20040830	AU	2003-303869	20030508
AU	2003269993	A1	20040311	AU	2003-269993	20030822

PRAI US 2002-227152 A 20020822
 US 2002-227195 A 20020822
 US 2001-280530P P 20010330
 US 2001-313264P P 20010817
 US 2001-166341 B2 20010918
 US 2001-323059P P 20010918
 US 2001-327006P P 20011005
 US 2001-332550P P 20011126
 US 2002-106097 A2 20020326
 US 2002-142364 A1 20020508
 WO 2003-US14799 W 20030508
 WO 2003-US26469 W 20030822

AB This invention relates to detection of **SNPs** and **haplotype** blocks within human chromosome 21 and assocn. with Long QT Syndrome (LQTS) and hearing disorders. Linkage studies are discussed, evaluating **SNPs** in the potassium channel genes KCNE1 and KCNE2, found on human chromosome 21 and assocd. with LQTS and hearing disorders. The **SNPs** can be used as susceptibility biomarkers in diagnosis of LQTS and hearing **diseases**, as well as predicting **drug** sensitivity in LQTS patients.

L3 ANSWER 36 OF 41 MEDLINE on STN DUPLICATE 12
Full Text
 AN 2004190601 MEDLINE
 DN PubMed ID: 15088270
 TI A high-density admixture map for **disease** gene discovery in african americans.
 AU Smith Michael W; Patterson Nick; Lautenberger James A; Truelove Ann L; McDonald Gavin J; Waliszewska Alicja; Kessing Bailey D; Malasky Michael J; Scafe Charles; Le Ernest; De Jager Philip L; Mignault Andre A; Yi Zeng; De The Guy; Essex Myron; Sankale Jean-Louis; Moore Jason H; Poku Kwabena; Phair John P; Goedert James J; Vlahov David; Williams Scott M; Tishkoff Sarah A; Winkler Cheryl A; De La Vega Francisco M; Woodage Trevor; Sninsky John J; Hafler David A; Altshuler David; Gilbert Dennis A; O'Brien Stephen J; Reich David
 CS Laboratory of Genomic Diversity, National Cancer Institute, Frederick, MD, USA.
 NC HL-65234 (United States NHLBI)
 K-01 HG002758-01 (United States NHGRI)
 K08 NS046341 (United States NINDS)
 N01-C0-12400
 U19-AI50864 (United States NIAID)
 SO American journal of human genetics, (2004 May) Vol. 74, No. 5, pp. 1001-13. Electronic Publication: 2004-04-14.
 Journal code: 0370475. ISSN: 0002-9297.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 (RESEARCH SUPPORT, NON-U.S. GOV'T)
 (RESEARCH SUPPORT, U.S. GOV'T, P.H.S.)
 LA English
 FS Priority Journals
 EM 200406
 ED Entered STN: 17 Apr 2004
 Last Updated on STN: 4 Jun 2004
 Entered Medline: 3 Jun 2004
 AB Admixture mapping (also known as "mapping by admixture linkage disequilibrium," or MALD) provides a way of localizing genes that cause **disease**, in admixed ethnic groups such as African Americans, with approximately 100 times fewer markers than are required for whole-genome **haplotype scans**. However, it has not been possible to perform powerful **scans** with admixture mapping because the method requires a dense map of validated markers known to have large frequency differences between Europeans and Africans. To create such a map, we screened through **databases** containing approximately 450000 single-nucleotide

polymorphisms (**SNPs**) for which frequencies had been estimated in African and European population samples. We experimentally confirmed the frequencies of the most promising **SNPs** in a multiethnic panel of unrelated samples and identified 3011 as a MALD map (1.2 cM average spacing). We estimate that this map is approximately 70% informative in differentiating African versus European origins of chromosomal segments. This map provides a practical and powerful tool, which is freely available without restriction, for screening for **disease** genes in African American patient cohorts. The map is especially appropriate for those **diseases** that differ in incidence between the parental African and European populations.

L3 ANSWER 37 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2003:511479 CAPLUS

DN 139:80261

TI Polymorphic sites in human metabotropic glutamate receptor mGluR8 gene and diagnostic and therapeutic uses thereof

IN Hess, John W.; Warren, Lee Evan; Conn, Jeffrey

PA Merck & Co., Inc., USA

SO PCT Int. Appl., 109 pp.

CODEN: PIXXD2

DT Patent

LA English

FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	WO 2003054167	A2	20030703	WO 2002-US41294	20021219
	WO 2003054167	A3	20031218		
	W: CA, US				
	RW: AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, SI, SK, TR				
	CA 2471198	A1	20030703	CA 2002-2471198	20021219
	EP 1456418	A2	20040915	EP 2002-805679	20021219
	R: AT, BE, CH, DE, DK, ES, FR, GB, GR, IT, LI, LU, NL, SE, MC, PT, IE, SI, FI, CY, TR, BG, CZ, EE, SK				
	US 2005233321	A1	20051020	US 2004-499580	20040621
PRAI	US 2001-342555P	P	20011220		
	WO 2002-US41294	W	20021219		

AB This invention relates to polymorphisms in the human metabotropic glutamate receptor mGluR8 gene, GRM8, and in particular to the discovery of 10 single nucleotide polymorphisms in the mGluR8 gene. Five polymorphic sites (PS1-PS3, PS6, and PS9) are located in coding exons, one (PS10) affects RNA splicing and results in a truncated polypeptide, and four polymorphic sites (PS4, PS5, PS7, and PS8) are located in introns. The invention also relates to methods and materials for analyzing allelic variation in the mGluR8 gene, and to the use of mGluR8 polymorphisms in the diagnosis and treatment of mGluR8 and/or mGluR8-mediated **diseases**, such as Parkinson's **disease** etc. The herein disclosed probes contg. at least one of the herein disclosed **SNPs** can be used to **identify** nucleic acid samples contg. mGluR8 **SNPs** or as primers or for expressing variant proteins. The invention includes methods of analyzing the polymorphic forms occupying the polymorphic sites. The invention further claims use of genotypes of the mGluR8 **SNPs** in treatment of humans with mGluR8 receptor antagonist **drugs**. In addn., the invention claims a method for screening therapeutic mGluR8 agonists or antagonists by their effects on human mGluR8 bioactivity or interactions with binding partners.

L3 ANSWER 38 OF 41

MEDLINE on STN

DUPLICATE 13

Full Text

AN 2003281292 MEDLINE

DN PubMed ID: 12808430

TI Identification of a high-risk **haplotype** for the dystrobrevin binding protein 1 (DTNBP1) gene in the Irish study of high-density schizophrenia families.

AU van den Oord E J C G; Sullivan P F; Jiang Y; Walsh D; O'Neill F A; Kendler K S; Riley B P

CS Department of Psychiatry and Virginia Institute for Psychiatric and Behavioral Genetics, Medical College of Virginia, Virginia Commonwealth University, Richmond, VA 23298-0126, USA.. eivandenoord@vcu.edu

NC MH41953 (United States NIMH)

MH45390 (United States NIMH)

MH52537 (United States NIMH)
SO Molecular psychiatry, (2003 May) Vol. 8, No. 5, pp. 499-510.
Journal code: 9607835. ISSN: 1359-4184.
CY England: United Kingdom
DT Journal; Article; (JOURNAL ARTICLE)
(RESEARCH SUPPORT, U.S. GOV'T, P.H.S.)
LA English
FS Priority Journals
EM 200403
ED Entered STN: 17 Jun 2003
Last Updated on STN: 12 Mar 2004
Entered Medline: 11 Mar 2004
AB A recent report showed significant associations between several **SNPs** in a previously unknown EST cluster with schizophrenia. (1). The cluster was identified as the human dystrobrevin binding protein 1 gene (DTNBP1) by sequence **database** comparisons and homology with mouse DTNBP1. (2). However, the linkage disequilibrium (LD) among the **SNPs** in DTNBP1 as well as the pattern of significant **SNP**-schizophrenia association was complex. This raised several questions such as the number of susceptibility alleles that may be involved and the size of the region where the actual **disease** mutation(s) could be located. To address these questions, we performed different single-marker tests on the 12 previously studied and 2 new **SNPs** in DTNBP1 that were re-scored using an improved procedure, and performed a variety of **haplotype** analyses. The sample consisted of 268 Irish multiplex families selected for high density of schizophrenia. Results suggested a simple structure where the LD in the **target** region could be explained by 6 **haplotypes** that together accounted for 96% of **haplotype** diversity in the whole sample. From these six, a single high-risk **haplotype** was identified that showed a significant association with schizophrenia and explained the pattern of significant findings in the analyses with individual markers. This **haplotype** was 30 kb long, had a large effect, could be measured with two tag **SNPs** only, had a frequency of 6% in our sample, seemed to be of relatively recent origin in evolutionary terms, and was equally distributed over Ireland. Implications of these findings for follow-up and replication studies are discussed.

L3 ANSWER 39 OF 41 MEDLINE on STN DUPLICATE 14
Full Text
AN 2003063510 MEDLINE
DN PubMed ID: 12573264
TI Single-nucleotide polymorphisms in the Toll-like receptor 9 gene (TLR9): frequencies, pairwise linkage disequilibrium, and **haplotypes** in three U.S. ethnic groups and exploratory case-control **disease** association studies.
AU Lazarus Ross; Klimecki Walter T; Raby Benjamin A; Vercelli Donata; Palmer Lyle J; Kwiatkowski David J; Silverman Edwin K; Martinez Fernando; Weiss Scott T
CS Channing Laboratory, Department of Medicine, Brigham and Women's Hospital and Harvard Medical School, Boston, MA 02115, USA..
ross.lazarus@channing.harvard.edu
NC U01 HL66795 (United States NHLBI)
SO Genomics, (2003 Jan) Vol. 81, No. 1, pp. 85-91.
Journal code: 8800135. ISSN: 0888-7543.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
(RESEARCH SUPPORT, U.S. GOV'T, P.H.S.)
LA English
FS Priority Journals
EM 200309
ED Entered STN: 8 Feb 2003
Last Updated on STN: 6 Sep 2003
Entered Medline: 5 Sep 2003
AB TLR9 is a mammalian Toll-like receptor homologue that appears to function as an innate immune pattern recognition protein for motifs that are far more common in bacterial than in mammalian DNA. The gene was sequenced in 71 subjects from three self-identified U.S. ethnic groups to **identify** single-nucleotide polymorphisms (**SNPs**). A total of 20 **SNPs** were found of which only 20% were in the public dbSNP **database**. Four **SNPs** were relatively common in all three ethnic samples. Using these four **SNPs**, seven distinct **haplotypes** were statistically inferred, of which four accounted for 75% or more chromosomes. These four **haplotypes** could be

distinguished from each other by the alleles of two **SNPs** (-1237 and 2848). Five exploratory nested case-control **disease**-association studies (asthma, DVT, MI, and COPD in European Americans and asthma in African Americans) were performed by genotyping DNA collected from four ongoing cohort studies. There was evidence suggesting increased risk for asthma with a C allele at -1237 (odds ratio 1.85, 95%CI 1.05 to 3.25) among European Americans (genotypes available from 67 cases and 152 controls). No other significant **disease** associations were detected. Replication of this finding in other, larger samples is needed. This study suggests that there is substantial diversity in human TLR9, possibly associated with asthma in Europeans but not African Americans. No association was detected with three other **diseases** potentially related to innate immunity.

L3 ANSWER 40 OF 41 CAPLUS COPYRIGHT 2008 ACS on STN

Full Text

AN 2003:602483 CAPLUS

DN 139:259338

TI Independent mutational events are rare in the ATM gene: **haplotype** prescreening enhances mutation detection rate

AU Mitui, Midori; Campbell, Catarina; Coutinho, Gabriela; Sun, Xia; Lai, Chih-Hung; Thorstenson, Yvonne; Castellvi-Bel, Sergi; Fernandez, Luis; Monros, Eugenia; Carvalho, Beatriz Tavares Costa; Porras, Oscar; Fontan, Gumersindo; Gatti, Richard A.

CS Department of Pathology and Laboratory Medicine, The David Geffen School of Medicine, Los Angeles, CA, USA

SO Human Mutation (2003), 22(1), 43-50

CODEN: HUMUE3; ISSN: 1059-7794

PB Wiley-Liss, Inc.

DT Journal

LA English

AB Mutations in the ATM gene are responsible for the autosomal recessive disorder ataxia-telangiectasia (A-T). Many different mutations have been identified using various techniques, with detection efficiencies ranging from 57 to 85%. In this study, we employed short tandem repeat (STR) **haplotypes** to enhance mutation identification in 55 unrelated A-T families of Iberian origin (20 Spanish, 17 Brazilian, and 18 Hispanic-American); we were able to **identify** 95% of the expected mutations. Allelic sizes were standardized based on a ref. sample (CEPH 1347-2). Subsequent mutation screening was performed by PTT, SSCP, and DHPLC, and abnormal regions were sequenced. Many STR **haplotypes** were found within each population and six **haplotypes** were obsd. across several of these populations. Single nucleotide polymorphism (**SNP**) **haplotypes** further suggested that most of these common mutations are ancestrally related, and not hot spots. However, two mutations (8977C>T and 8264_8268delATAAG) may indeed be recurring mutational events. Common **haplotypes** were present in 13 of 20 Spanish A-T families (65%), in 11 of 17 Brazilian A-T families (65%), and, in contrast, in only eight of 18 Hispanic-American families (44%). Three mutations were identified that would be missed by conventional screening strategies. In all, 62 different mutations (28 not previously reported) were identified and their assocd. **haplotypes** defined, thereby establishing a new **database** for Iberian A-T families, and extending the spectrum of worldwide ATM mutations.

RE.CNT 33 THERE ARE 33 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 41 OF 41 MEDLINE on STN

Full Text

AN 2002729804 MEDLINE

DN PubMed ID: 12491775

TI **SNP database** and establishment of personalized medicine.

AU Inoue Ituro

CS Division of Genetic Diagnosis, Institute of Medical Science, University of Tokyo, 4-6-1 Shirokanedai, Minato-ku, Tokyo 108-8639, Japan.

SO Nippon yakurigaku zasshi. Folia pharmacologica Japonica, (2002 Nov) Vol. 120, No. 1, pp. 41P-42P. Ref: 0

Journal code: 0420550. ISSN: 0015-5691.

CY Japan

DT (ENGLISH ABSTRACT)

Journal; Article; (JOURNAL ARTICLE)

General Review; (REVIEW)

LA Japanese

FS Priority Journals
 EM 200304
 ED Entered STN: 21 Dec 2002
 Last Updated on STN: 2 Apr 2003
 Entered Medline: 1 Apr 2003
 AB We are aiming to **identify** susceptibility genes for common or otherwise clinically relevant **diseases** of metabolism such as diabetes, asthma, and hypertension, and analyze the molecular causality. Although genetic and environmental factors play equally crucial roles in the pathogenesis of the common **diseases** of civilization, genetic factor is directly involved in the causality and molecular mechanism. The elucidation of molecular etiology provides specific molecular **targets** for therapeutic **drugs** even at the individual level. Thus our priority is analysis of the molecular causality of the common metabolic disorders of civilization. We will **identify** individual and group polymorphisms (**SNPs**) in the genome relevant to the treatment of individual patients closely related to susceptibility to **disease**, prognosis of **disease**, and responses to **drugs**. To determine the genetic susceptibilities, we apply genetic approaches such as linkage studies with affected sib-pairs and association studies using **SNPs database** together with **haplotype** analysis.

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CA SUBSCRIBER PRICE	-10.40	-10.40

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